

PSYCHOMETRIC TESTING OF A SCALE DESIGNED TO
MONITOR THE PSYCHOSOCIAL AND BEHAVIORAL
IMPACT OF GENETIC TESTING FOR HEREDITARY
NONPOLYPOSIS COLORECTAL CANCER (HNPCC):
A PILOT STUDY

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**Psychometric Testing of a Scale Designed to Monitor the
Psychosocial and Behavioral Impact of Genetic Testing for
Hereditary Nonpolyposis Colorectal Cancer (HNPCC): A Pilot Study**

by

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Abstract

The purpose of this pilot study was to conduct preliminary psychometric testing on the Hereditary Diseases and Genetic Testing (HD-GT) scale, which was designed to monitor the psychosocial and behavioral impact of genetic testing for hereditary nonpolyposis colorectal cancer (HNPCC) or Lynch syndrome. The pilot was part of a larger study which is using a descriptive correlational design with longitudinal components to develop, validate and evaluate monitoring tools for individuals with genetic-based diseases. The framework for this study was the substantive theory, *Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases* (Way et al., 2008).

The target population was individuals at 50% risk for inheriting HNPCC who had participated in genetic testing and were informed of their carrier status. Survey respondents were recruited from population-based probands comprising the Provincial Medical Genetics Program of Newfoundland and Labrador. Study participants (N = 75) were similar to the target population in terms of personal and illness-related characteristics. Data were collected by face-to-face interviews, telephone interviews and self-administered surveys between February and May 2008.

Psychometric testing of the HD-GT scale was based on the work of Ware and Gandek (1998). Preliminary findings are indicative of good data quality and potential usability of the scale under variant administrative conditions. As well, all of the HD-GT subscales met the criteria for Likert scaling assumptions and evidence very good reliability and validity.

The various subscales of the HD-GT augment what has been reported in the literature and also provide new insights into the psychosocial and behavioral impact of genetic testing for individuals and families with HNPCC. Study findings suggest that the family history of cancer does have a significant impact on decision-making regarding genetic testing. There are also indications that study respondents place high value on having all potentially at-risk family members participate in genetic testing, but are often challenged trying to convince them to accept the need for testing.

With regard to the genetic testing process, most respondents placed high value on being emotionally prepared for genetic testing and having appropriate information, but not everyone required health care provider or family/friends support. As well, despite experiencing some emotional difficulty while waiting for test results, not everyone required support prior to and during the receipt of results. Most respondents, however, do place high value on receiving a follow-up letter to reinforce their genetic testing results.

Most respondents understood the importance attached to being proactive in leading a healthy life and participating in cancer screening. They also believed that there was an increased cancer presence among young people in the family. Finally, most family members, young and old alike, wanted information about HNPCC, and were perceived to understand it, but encountered some difficulties in communicating the information to other family members.

Study findings indicate that the subscales appear to be sensitive enough to measure the wide-range of psychosocial and behavioral implications of genetic testing. However, due to study limitations, generalizability of the findings is cautioned until the larger study is complete. The findings also provide support for previous research and suggest that

more research is needed to inform the practice of genetic counseling. There is also a need for further research into the psychological implications of having an inconclusive test result.

Finally, study findings have important implications for nursing practice.

Competencies required for the effective delivery of genetic services need to be built into the scope of professional nursing practice. If nurses are to work effectively with HNPCC families they must have the appropriate knowledge, education and skills to recognize the features of HNPCC, to take thorough patient and family histories, to provide support, and to coordinate care for these individuals.

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CHAPTER 1

Introduction

Advances in DNA technology have made it possible to predict the risk status of individuals with an inherited predisposition for certain conditions (Reeve, Owens, & Winship, 2000). In the early 1990's genetic testing became available for several cancer types (Bleiker, Hahn, & Aaronson, 2003). Today, it is a common and integral part of cancer services in many countries. Health care providers are ethically responsible for ensuring that the information provided about genetic predisposition risks is not detrimental to the psychological well-being of individuals and their families. Importantly, the information conveyed should be well understood by the intended recipient and reference recommended screening and surveillance protocols (Collins et al., 2007).

Motivational factors behind the decision to have genetic testing are complex and varied (Reeve et al., 2000). It seems that when treatment options are limited (such as with Huntington's disease), genetic testing uptake is much lower with many at-risk individuals not wanting to know their status. A contrasting perspective is evident for diseases, like colorectal cancer (CRC), which can be effectively treated when diagnosed early. Some jurisdictions report a genetic testing uptake of approximately 75% for CRC (Bleiker et al., 2003).

Conspicuously limited in the genetics literature are research findings on how awareness and understanding of a confirmed familial hereditary nonpolyposis colorectal cancer (HNPCC) or Lynch syndrome presence influences decision-making prior to and following genetic testing. As well, we have limited insight into the implications of

participating in genetic testing for HNPCC for individuals in affected families.

Surprisingly, research findings have documented minimal psychological and behavioral impacts (Claes et al., 2005; Collins et al., 2007; Heshka, Palleschi, Howley, Wilson, & Wells, 2008). Certain individuals may be burdened by knowing that they are at increased risk for a life-threatening disease, while those testing negative for the gene mutation may be relieved (Bleiker et al., 2003). Other individuals may be given inconclusive results (non-confirmed carrier or non-carrier status) and thus must live with continuing uncertainty about their risk status (Lindor et al., 2006). Finally, we have limited understanding of how families communicate relevant information to their members about their potential risks (Gaff, Collins, Symes, & Halliday, 2005; Lim et al., 2004; Mesters, Ausems, Eichhorn, & Vasen, 2005; Peterson et al., 2003).

Another significant gap in the genetics literature is information on the motivational factors responsible for behavioral change in concordance with recommended screening (Bleiker et al., 2003; Marteau & Weinman, 2006; McAllister, 2002, 2003; Meiser, 2005; Murakami et al., 2004). Some evidence suggests that the worry and upset felt by carriers may deter participation in regular screening which is crucial for prevention and early detection (Codori, Petersen, Miglioretti, & Boyd, 2001; Jarvinen, Mecklin, & Sistonen, 1995; Lerman, Marshall, Audrain, & Gomez-Caminero, 1996).

Several authors emphasize the need for more research in the area, with some advocating for incorporating qualitative methods into study designs (Bleiker et al., 2003; Marteau & Weinman, 2006; McAllister, 2001; Riper, 2005). Greater interest in qualitative approaches over the past decade has emerged in response to the increased recognition given to the role played by psychosocial factors in shaping health outcomes

(Gregory & Way, 2008). Certainly further research is needed to increase health care providers' and policy makers' understandings of the impact of genetic testing on individuals and families, and to use this information in planning for and implementing appropriate services (Aktan-Collan, Meekin, & Kaariainen, 2001a; Aktan-Collan, Haukkala, Mecklin, Uutela, & Kaarianen, 2001b; Bleiker et al., 2003; Gaff et al., 2005; Hamilton, Bowers, & Williams, 2005; Jarvinen et al., 2000; Mesters et al., 2005; Tiller et al., 2005).

The purpose of the current study is to provide support for what has been reported in the literature to date, as well as to provide new insights regarding the psychosocial and behavioral impact of genetic testing for HNPCC. A qualitative database was used as the foundation to develop an instrument for use in the pre- and post-genetic testing phases, as well as during the process of genetic testing. It is believed that the use of a qualitative database will result in an instrument that is sensitive enough to measure the full psychosocial and behavioral impact of HNPCC. This instrument is being tested in the Newfoundland and Labrador (NL) HNPCC population.

Background and Rationale

HNPCC is the most common form of hereditary CRC (Lynch & de la Chapelle, 2003). It is an autosomal dominant disorder with a germline mismatch repair (MMR) mutation segregating within high and intermediate risk families. Although conjectured to be responsible for only 1% to 3% of all CRC cases world-wide, the situation in Newfoundland and Labrador (NL) is quite different with estimates of up to 47% of all individuals with CRC coming from families at high or intermediate risk for HNPCC

(Stuckless et al., 2007). The high prevalence of genetic based cancers in the province has significant implications for health policy directed toward improving the overall health of the population.

HNPCC and Genetic Testing

Microsatellite instability (MSI) is the genetic basis for tumor development in HNPCC. Mutations occurring in microsatellites during replication are normally repaired by MMR proteins. When this mechanism is impaired or deficient, as with MSI, errors are not corrected (Kouraklis & Misiakos, 2005).

Germline mutations may occur in one of several MMR genes. The most common genes are MSH2, MLH1, PMS1 and PMS2, with MSH2 and MLH1 accounting for 60% and 30% of HNPCC cases, respectively. Significantly, extracolonic carcinomas tend to occur more frequently in families with the MSH2 mutation (Stuckless et al., 2007). Other newer and less common mutations include MSH6 which is responsible for a more atypical and benign form of HNPCC, and MLH3 which is present in only a small percent of HNPCC-like families (Kouraklis & Misiakos, 2005). In NL, three MSH2 gene mutations have been identified: exon 8 deletion, exon 4-16 deletion and intron 5 splice mutation resulting in deletion of exon 5 in mRNA (Stuckless et al.).

There are several cardinal features of HNPCC that geneticists use to determine the high or intermediate risk status of families. Some of the more important clinical profiles include: early average age of cancer onset (i.e., early to mid-40s and lower); proximal colon cancer involvement (70% of cases); accelerated carcinogenesis or decreased time line between first detection of a tiny adenoma and its progression to carcinoma (i.e.,

within 2 to 3 years); increased probability for new colon primaries (i.e., about 25% to 30% within 10 years of surgical resection); and greater risk for extracolonic carcinomas (especially the endometrium and to lesser degrees the ovary, stomach, small bowel, hepatobiliary tract, pancreas, upper uro-epithelial tract, and brain) (Lynch & de la Chapelle, 2003; Lynch, Lynch, Lynch, & Attard, 2008; Merg, Lynch, Lynch, & Howe, 2005).

Genetic testing for HNPCC mutations should be offered to individuals who have a high probability of developing CRC and related cancers based on personal and family histories of the disease. This is especially true for familial cancer profiles that meet the Amsterdam or modified Amsterdam criteria and/or Bethesda guidelines (Halbert et al., 2004). The reader is referred to Appendix A for a summary of these guidelines.

If DNA testing reveals that a family member is a HNPCC carrier, lifetime cancer risk rises to approximately 90%. Conversely, if a family member is not a carrier, the cancer risk returns to that of the general population (Chung & Rustgi, 2003). Endometrial and ovarian cancers occur in up to 60% and 10%, respectively, of female mutation carriers. Gastric and urothelial cancers also carry a lifetime risk of about 10%. Other cancers, like the small bowel and biliary system, occur in much smaller proportions in HNPCC families (Aarnio et al., 1999; Dunlop et al., 1997; Stuckless et al., 2007).

The type and time of cancer onset for HNPCC carriers varies across individuals and families. Lynch syndrome shows incomplete penetrance (not all mutation carriers will develop a cancer) and variable expressivity (individuals develop different cancers at different ages). The relationship between genotype (gene level mutations) and phenotype (physical characteristics) in HNPCC is not well understood, despite the identification of a

large number of predisposing mutations. Stuckless et al. (2007) reported that the cumulative risk of developing cancer by age 70 is 94% in carriers of the MSH2 mutation in the NL population. This presents significant implications for the current study which is focused on this population.

Another equally important consideration is the limited diagnostic utility of current DNA technologies. Despite great advances and the concomitant benefits for families with Lynch syndrome-like profiles, a MMR mutation is only identified in about one-half of these families. This may be due to the inability of conventional technologies to detect alterations in suspected genes or to identify new genes that are the responsible agents (Lynch et al., 2008; Merg et al., 2005). The other problem is that a significant proportion of families presumed to be mutation negative may have hidden alternations in known predisposition genes (Lindor et al., 2006; Lynch et al., 2008). Because predictive DNA testing is not an exact science, a significant number of families must contend with inconclusive results. This reality has significant implications for health care providers.

Psychosocial and Behavioral Implications of Genetic Testing

Previous studies have been inconsistent in their ability to predict short- and long-term outcomes from participating in genetic testing (McAllister, 2002). Little is known about how families define themselves within the context of hereditary cancer, how they communicate cancer risk information, and what influences individual members' decisions to undergo genetic counseling and testing (Peterson et al., 2003). The presence of hereditary cancers has been found to exert variable effects on communication patterns within families (Gaff et al., 2005; Koehly et al., 2003; Mesters et al., 2005; Peterson et

al.) and family relations (Koehly et al.). There is also some empirical support for the conjecture that the family context can significantly influence members' decisions regarding participation in genetic testing (d'Agincourt-Canning, 2005; Koehly et al.; McAllister, 2002, 2003; Mesters et al.).

To date, limited research has been focused on evaluating the effectiveness of genetic counseling sessions in helping individuals make informed decisions about genetic testing, in preparing them for the process and in helping reduce the impact of this technology. The evidence is conflicting on the positive effect of genetic counseling sessions on cognitive and affective outcomes (Braithwaite, Emery, Walter, Prevost, & Sutton, 2004; McAllister, 2003). McAllister proposed that the attitudes and motivation of individuals towards genetic testing influence the degree of engagement in the process and, ultimately, variations in reactions post-testing. Despite the absence of consensus on the type and amount of information needed by individuals (Bleiker et al., 2003), some could likely benefit from ongoing contact with genetic counselors (Collins et al., 2007).

Frequent screening for CRC and other HNPCC-related cancers is the norm for individuals who are carriers of the HNPCC gene. Cancer surveillance, including colonoscopy, has to be conducted for a lifetime, which may be distressing to many individuals and also carries the risk of complications (Lackner & Hoefler, 2005). Prior to the advent of genetic testing in the 1990's, all individuals in HNPCC families were recommended to be screened based on their family histories (Halbert et al., 2004). This relatively new technology may lessen the burden of screening as those who test negative are not required to continue the vigorous screening schedule of their carrier relatives.

The identified knowledge gaps in the literature are important for framing the context of genetic counseling. Furthermore, genetic counselors have an obligation to adapt sessions to the needs and preferences of targeted individuals (Pieterse et al., 2005). To date, there is limited evidence to suggest that genetic counseling is based on evidence-based theoretical frameworks. The current study was conducted with the intention of helping to close this gap.

Problem Statement

A qualitative study previously conducted by the research team as part of the larger study found similar and different results from that documented in relevant literature. It was apparent from the interviews that individuals living within HNPCC families experience a wide range of emotional, psychological, social and physical issues that have important implications for their health and quality of life. These experiences were collapsed into three major thematic categories: living in families with a strong history of hereditary cancer; becoming aware of genetic testing and living the process; and struggling to adjust (with a positive/negative test result). The data from this study was used to generate a substantive theory, *Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases* (Way et al., 2008).

Following generation of the substantive theory, the research team used the operational indicators comprising the descriptors of each property defining each category to draft two scales – one focusing on the genetic testing process and the other on psychosocial and behavioral adjustment following genetic testing. The current pilot study was designed to conduct preliminary psychometric testing on one of these scales.

The Hereditary Diseases and Genetic Testing (HD-GT) scale is based on the first two themes from the qualitative study (living in families with a strong history of hereditary cancer; and becoming aware of genetic testing and living the process). The current study will add to the body of literature that addresses the influence of a family history of hereditary cancer on genetic testing decision-making, engagement in genetic testing, understanding and acceptance of cancer risk and recommended preventive actions, and communications about the genetic link to cancer with family members. The information generated by this scale has the potential to significantly influence the practice of genetic counseling for families with HNPCC and other genetic-linked diseases.

Purpose and Research Questions

The overall objective of this component of the project was to: a) test the feasibility of using the scale under variant conditions, b) validate subscale and overall scale structure and, c) examine scaling (rating) methods.

This pilot study was designed to address the following research questions:

1. Is it appropriate to administer the scale under variant conditions (i.e., telephone, self-administered, or face-to-face)?
2. How well do the various subscales of the HD-GT meet the Likert scaling assumptions?
3. Are the subscales reliable?
4. Are the subscales valid?

CHAPTER 2

Literature Review

The purpose of this review of the literature is to examine current research on the factors influencing uptake of predictive testing for HNPCC and the resulting impact on individuals and families. This review is framed within the context of the substantive theory, *Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases*, generated by the grounded theory phase of the larger study. The theory's major underlying premise is that familial contexts and individual psycho-emotional factors influence acceptance of the hereditary link to cancer, perception of risk, motivation to become involved in genetic testing, reaction to and understanding of test results, and willingness to share test results with others and discuss their potential risk (Way et al., 2008).

The first section of the literature review focuses on the psychological and emotional impacts of living in families with a strong history of cancer, and the reaction to and acceptance of a possible genetic link to the disease. The second section explores how variable levels of engagement in the genetic testing process may impact reactions to being informed about one's HNPCC status, decision-making concerning recommended behavioral changes and willingness to openly communicate about one's HNPCC status to immediate and extended family members. The final section presents an overview of the variant processes involved in developing monitoring tools for use in clinical settings.

Familial Cancer and the Genetic Link

It is estimated that close to 1000 families worldwide have the gene mutation that is responsible for HNPCC. Many individuals in these families have negative experiences related to losing relatives to cancer at a young age, caring for family members with cancer, and worrying about cancer risk for the self as well as one's siblings and children (Carlsson & Nilbert, 2007). Despite the increased identification of families with HNPCC, limited documentation exists on the psychosocial and emotional burden of living with an uncertain or evolving disease state, and short- and long-term quality outcomes. Further, the available data is limited on the factors influencing individuals' decision to have genetic testing and their motivation to follow recommended screening protocols with or without genetic testing.

Family Context and Risk Perception

Relatively few studies have examined the role played by the familial cancer context in shaping perceptions of personal risk or implications for siblings and offspring. Significantly, little is known as well about how families define themselves within the context of hereditary cancer, how members deal with personal and others' cancer experiences, and how individuals make the decision to undergo genetic counseling and testing (Mesters et al., 2005; Peterson et al., 2003).

The available evidence suggests that experiences with familial cancer and the resulting understandings about one's risk influence an individual's decision-making prior to and following genetic testing for HNPCC (Bleiker et al., 2003; Marteau & Weinman, 2006; McAllister, 2002, 2003; Meiser, 2005; Murakami et al., 2004). The most

enlightening insights into the importance of the family context for shaping risk perceptions, and how individuals prepare for and become involved in the process are provided by qualitative studies (d' Agincourt-Canning, 2005; McAllister, 2002, 2003). What these two researchers have in common is the priority given to experiences with close and distant relatives and the personal interpretations of these experiences in shaping perceptions of risk.

In a qualitative study of women's experiences with genetic testing for hereditary breast and ovarian cancer, d' Agincourt-Canning (2005) explored the connection between experiential knowledge gained from living with a family history of these diseases and personal understandings of cancer risk. Thematic analysis yielded two major categories of experiential knowing – empathetic (cancer knowledge constructed from family communications about the high prevalence, or exposure to disease progression and treatment responses in others, or caring for affected others) and embodied (personal physical and emotional experiences with cancer and its treatment). Both of these ways of knowing are purported to be used in constructing theories of cancer risk for the self and others (sense of vulnerability and/or perceived threat). This author concludes that the strong impact of personal experiences of cancer within the self or with others on perceptions of risk highlights its importance as a focus for genetic counselors to help better prepare individuals for genetic testing.

McAllister (2002, 2003) used a grounded theory approach to generate a Theory of Engagement which proposes that variable levels of cognitive and emotional involvement in the family cancer context are key factors influencing conceptualizations of risk about HNPCC and, ultimately, intensity of engagement in the genetic testing process. It is also

conjectured that the engagement process is a psychosocial one influenced by discussions about the familial cancer history, personal theories of inheritance (lay theories), coping styles and social factors (i.e., degree of personal experience with cancer in the self or other family members). The theory also proposes that the engagement process continuously evolves along a continuum from disengagement through partial (cognitive processing only) to intense (cognitive and emotional processing) engagement.

Genetic Testing Decision-Making

Individuals in HNPCC families seek genetic counseling for a number of reasons. Families play a key role in influencing individual members' awareness of cancer risk and decisions about genetic testing (Bleiker et al., 2003). Common motives for accessing genetic counseling services include a desire to obtain information about genetic testing, to discuss the family history, to get information on early detection and prevention, to obtain emotional support, and/or to reduce worry (Pieterse et al., 2005). Additional motivating factors include wanting to avoid unnecessary screening, to clarify one's status for the benefit of offspring and/or to inform reproductive decisions (Bleiker et al.; Carlsson & Nilbert, 2007; Claes et al., 2005; McAllister, 2002).

Research studies have been designed to quantify the impact of psychosocial and personal characteristics on genetic testing uptake. Study findings suggest that there tends to be a greater likelihood of genetic testing uptake in HNPCC families in the presence of personal experiences with cancer and/or exposure to a large number of affected relatives (Hadley et al., 2003) and greater perceived cancer risk or more frequent thoughts about cancer (Codori et al., 1999). Other researchers have found support for the positive impact

of an absence of depressive symptoms (Lerman et al., 1999), higher education (Lerman et al.) and active employment (Aktan-Collan et al., 2000) on genetic testing uptake.

Following a critical review of literature on psychosocial issues in cancer genetics, Bleiker et al. (2003) reported that perceived risks for cancer, more so than objective risks, are important influencers of genetic testing uptake. What this means is that if perceived susceptibility is high, individuals may seek genetic testing to decrease uncertainty, to confirm/refute the need to engage in preventive actions, to determine risk for children or to engage in future/family planning. These authors also noted that the research findings suggest that individuals seeking genetic testing tend to overestimate their risk and do not recall risk information very well. Furthermore, standardized education and counseling programs appear to have limited impact on modifying subjective risk perceptions. Finally, the evidence also suggests that high levels of distress and poor coping styles may impede the processing of risk information.

Studies conducted after Bleiker et al.'s (2003) review reinforce some of these findings. In a cross-sectional survey of 130 individuals attending a cancer clinic, Balmana, Stoffel, Emmons, Garber, and Syngal (2004) used a researcher-developed instrument to investigate the motivations and concerns of individuals prior to genetic counseling and testing for different hereditary cancer syndromes. Significant among the study findings was that most participants entered the process expecting a positive test result for the targeted syndrome and believed that they were at high to very high risk for cancer. As well, genetic testing seemed to be an enabling force behind decision-making about cancer prevention and medical management. Further, respondents believed that genetic testing would influence their future planning and their children's lives.

In a prospective study of 121 individuals with a family history of breast and colon cancer, Carlsson, Bjorvatn, Engebretsen, Berglund, and Natvig (2004) used standardized scales to assess cancer distress levels, self-efficacy, perceived social support and health-related quality of life. The findings in the pre-genetic counseling period indicated that one-fifth of the sample had significant distress scores. As well, less cancer-related distress and greater self-efficacy and perceived social support were found to be significantly associated with better mental health.

Summary

Currently, there is limited insight into the role played by the family context in shaping individuals' perceptions of HNPCC, including how they define their risk, make decisions about genetic counseling and testing, and communicate relevant information/experiences to other potentially at-risk family members. It has been conjectured that personal interpretations of experiences with familial cancer may shape perceptions of risk and influence engagement in the genetic testing process. With the available evidence suggesting that perceived cancer risk has a greater impact on genetic testing uptake than objective risk, genetic counselors should focus on developing greater insight into personal theories of risk. In short, more research is needed to understand the importance of the familial cancer context for shaping risk perception and genetic testing decision-making.

Genetic Testing Process

Collins et al. (2007) maintains that it is an ethical requirement of counseling programs for genetic testing to ensure that the information provided is understood by patients. The review of literature for the current study found few investigations that focused on evaluating how effective genetic counseling sessions are in preparing individuals for or reducing the impact of genetic testing. There also seems to be limited consensus on optimal counseling programs for ensuring that individuals make informed decisions and experience minimal negative impacts from this new technology.

Some authors argue that there is no evidence to support the premise that genetic counseling sessions increase the knowledge base and understanding levels of individuals (McAllister, 2003). However, in a systematic review and meta-analysis of the genetic-based literature, Braithwaite et al. (2004) did find support for increased knowledge of cancer genetics following genetic counseling.

Involvement in the Process

McAllister (2002, 2003) proposed that the psychological concept of engagement influences attitudes and motivation towards genetic testing for HNPCC. McAllister (2003) conjectured that variations in degrees of engagement may help explain differences in how individuals approach and react to genetic testing. Engagement can be partial (cognitively aware of the 50:50 inheritance probability) or intense (fearful and anxious with strong convictions about one's carrier status), with disengagement occurring if the whole idea about the inheritability of cancer is unacceptable or too painful.

The research evidence suggests that many individuals with a family history of CRC experience high levels of perceived risk for developing the disease (Bleiker et al., 2003; Braithwaite et al., 2004; Croyle & Lerman, 1999). Elevated risk perceptions for cancer have been associated with psychological distress prior to, during and following genetic testing (Bleiker et al.; Esplen et al., 2003).

In an updated review of the literature, Meiser (2005) explored the psychological and behavioral impact of participating in the genetic testing process. One important finding not referenced previously was the limited information available on how the delays experienced by certain individuals while waiting for their test results could enhance psychological distress. The author highlights this as a significant gap because of the probability of increased frustration and distress when future decision-making about screening and prophylactic interventions is contingent upon such results.

Bleiker et al.'s (2003) review failed to detect evidence-based data on the professional service needs of individuals and families undergoing predictive genetic testing. These authors argued that more research is needed to determine the type and amount of information required pre- and post-testing, how to provide better understanding of genetic testing and its effects, and how to best facilitate risk-reducing behavior without increasing emotional distress or providing false reassurances. Individuals who should be targeted for greater guidance and support are "family messengers." Other individuals who may require greater attention include those who fall into the following categories: exposed to cancer in a family member at a young age and thus could be at greater risk for adverse emotional consequences; experienced high levels of distress prior to testing; experienced the loss of a close relative to cancer shortly before

testing; personal experience with cancer; refused to receive genetic testing results; and, experienced a discrepancy between expected and actual outcomes of genetic testing.

Pieterse et al. (2005) noted that the evidence suggests that it is important to identify the needs and concerns of individuals seeking genetic counseling in order to better support them, lessen their anxiety and discuss complex issues with them. These authors developed an instrument to measure the needs and preferences of individuals participating in genetic counseling for hereditary cancer. Study findings suggest that a major concern of individuals prior to counseling is wanting information about personal and family members risk, preventive strategies, and the genetic testing process.

Reaction to HNPCC Status

The consequences of genetic testing vary widely depending on the individual and family. Certain individuals may become burdened by knowing that they are at increased risk for developing a life-threatening disease (Bleiker et al., 2003). Other individuals may experience anxiety and worry, feel guilty about passing on the gene and experience uncertainty about future health states (Claes et al., 2005; Espen et al., 2003). Besides the psychological and emotional repercussions, there may be negative effects on family relationships, a loss of privacy and discrimination by insurance companies and employers (Bleiker et al.; Carlsson & Nilbert, 2007; Hadley et al., 2003).

Although a negative genetic test result should provide relief, the literature describes what is known as 'survivor's guilt' among persons not found to have the gene mutation for HNPCC (Bleiker et al., 2003). In addition, while the risk for non-carriers of HNPCC

is the same as that of the general population, these individuals may find it difficult to discontinue screening (Hadley et al., 2004).

Based on an extensive review of relevant literature, Bleiker et al. (2003) reported that the general conclusion of research studies and meta-analyses is that there are no adverse psychological consequences from engaging in predictive testing. These findings have been attributed, in part, to the impact of genetic counseling on reducing anxiety and improving accurate perceptions of risk. However, Bleiker et al. argue that the assertion that genetic counseling decreases psychological distress and increases knowledge/perception of risk is mostly based on speculation.

Braithwaite et al. (2004) conducted a systematic review and meta-analysis of controlled trials and prospective studies focusing on outcomes of genetic counseling for familial cancer. The analysis revealed conflicting findings for cognitive and affective outcomes. Genetic counseling was observed to exert a consistent positive effect on knowledge of cancer genetics in both controlled trials and prospective studies. In contrast, no significant effects were observed for either risk perceptions or risk accuracy in the controlled trials, but significant improvements in risk accuracy were reported for most prospective studies. Similarly, the controlled trials, for the most part, failed to document a significant effect for genetic counseling on levels of general anxiety, general distress, depression or cancer-specific worry. In contrast, prospective studies documented statistically significant reductions in general anxiety in the short-term, inconsistent findings concerning reductions in general distress, no significant impact on depression and significant short but not long-term reductions in cancer-specific worry.

Meiser (2005) reviewed the literature on the psychological impact of genetic testing for hereditary breast/ovarian cancer susceptibility, HNPCC and Familial Adenomatous Polyposis (FAP). This author found that HNPCC studies consistently reported that unaffected non-carriers experience benefits (short- and long-term decrease in colon cancer anxiety, generalized anxiety, and depression) and evidence no adverse long-term effects from having genetic testing. With regard to carriers, there were only temporary increases in generalized anxiety immediately following disclosure of test results. It was common to also see short- and medium-term decreases in depression. Meiser concluded that these results may reflect the demonstrated effectiveness of screening for HNPCC.

A final important point emerging from Meiser's (2005) review is that it is not always possible to identify the specific gene mutation from tumors taken from family members with cancer. In fact, gene mutations are only identified in 50% of families with aggregate cancers, thus leaving a fairly large number of individuals with inconclusive results. The challenges here are two-fold. First, the absence of a known gene mutation may create false assurances regarding cancer risks. Second, certain individuals may experience psycho-emotional difficulties with the uncertainty of inconclusive results.

Several studies have been conducted since these literature reviews of earlier studies. Prospective studies have collected baseline and follow-up data on individuals participating in genetic testing. These studies are increasing our understanding of the psychosocial, medical, and behavioral impact of testing for mutations that predispose to cancer (Claes et al., 2005; Collins et al., 2007; Espen et al., 2007).

Claes et al. (2005) assessed distress (cancer-specific, state anxiety, global dysfunctioning) and illness representations (risk perception, perceived severity, and

perceived controllability of disease) in a sample of carriers and non-carriers of HNPCC one year after disclosure of genetic test results. A second focus of the study was to identify the best predictors of lower distress levels. Study results confirmed previous findings that genetic testing for HNPCC does not induce major psychological problems. The authors speculated that this may be attributed to focusing on the benefits of regular screening for early detection of cancer.

A more recent study on the psychological impact of HNPCC testing found that cancer worry, anxiety and depression levels either increase or remain the same for healthy carriers immediately after notification of test results compared to non-carriers (Esplen et al., 2007). As well, despite the tendency for increased distress levels to return to baseline at one year post-disclosure of genetic testing results, there continues to be a significant group of individuals who have elevated psychological symptoms, especially young female survivors, non-whites, the less educated and those with less social support. Other factors that affect distress include coping style and experience with loss.

Collins et al. (2007) also studied the impact of genetic testing three years after the receipt of results for predictive testing for HNPCC (N = 114). The findings suggested that the carriers' distress levels at 3-years were similar to those observed at 12-months (N = 73). As well, depression levels were very low and had returned to baseline.

Heshka et al. (2008) conducted a systematic review of existing studies dealing with perceived risks and the psychological and behavioral impacts of participating in predictive DNA testing for various cancers, including HNPCC. The authors found that overall, genetic testing had no significant impact on psychological outcomes (general and specific distress, anxiety, or depression) in either carriers or non-carriers. The impact on

cancer worry was less conclusive due to the limited comparative research. In addition, no significant differences were observed in the risk perceptions of carriers and non-carriers at 12 months which was lower than at baseline in most studies. The authors concluded that genetic testing does not appear to have negative repercussions for carriers or non-carriers.

Decision-Making and Behavioral Change

One important potential benefit of genetic testing for HNPCC is being able to make informed decisions about cancer screening directed toward helping reduce the incidence of and mortality from CRC (Collins et al., 2007; Hadley et al., 2003; Reeve et al., 2000). However, it cannot be assumed that carriers will necessarily adopt appropriate screening and preventive behaviors following genetic testing (Meiser, 2005). It is argued that quantitative studies have not been consistent in their ability to predict outcomes from genetic testing (McAllister, 2002). The extent to which a condition is considered to be preventable is an important predictor of whether or not individuals engage in screening (Marteau & Croyle, 1998).

In a systematic review and meta-analysis of studies focusing on genetic testing for cancer, Braithwaite et al. (2004) only identified a few studies that addressed cancer surveillance practices, with most documenting minimal changes in behavior. According to Meiser (2005), the focus of existing literature has been largely on the psychological impact of genetic testing, thus more research is needed on the behavioral impact of this technology. In a systematic review of studies dealing with the behavioral impact of participating in predictive DNA testing, Heshka et al. (2008) found that, for the most part,

self-reported screening practices (e.g., mammography, transvaginal ultrasound, cancer antigen, colonoscopy, etc.) increased post-genetic testing for both groups but significantly more so for carriers.

In their follow-up study, Claes et al. (2005) also assessed the health-related behavior (colonoscopy and transvaginal ultrasound) of carriers and non-carriers of HNPCC one year post-genetic testing. The hypothesized higher subjective risk perceptions of carriers versus non-carriers were not confirmed, but carriers did have higher uptake of screening post-testing.

Bleiker et al. (2005) conducted a study of 149 individuals who had undergone genetic counseling for CRC and advised to undergo periodic screening because of familial CRC or HNPCC. Participants were invited to complete a self-report questionnaire on psychosocial issues and screening practices. Noncompliance with screening advice was rare (3%) but significant delays (more than 1 year) were reported by approximately 25% of respondents. Some of the identified barriers to screening included not receiving a reminder letter, the embarrassing nature and discomfort associated with the procedure, fear that a tumor would be detected, and absence of symptoms.

Collins et al. (2007) also investigated the screening practices of carriers and non-carriers three years after predictive testing for HNPCC. All confirmed carriers had a colonoscopy within three years. The finding that only 7% of non-carriers had a colonoscopy suggests that these individuals are reassured by a negative test result and are willing to discontinue screening. Noteworthy is that these findings contrast with those of

other studies which suggest that non-carriers might be reluctant to discontinue screening (Hadley et al., 2004).

Some research studies have focused on identifying possible factors responsible for screening uptake. The evidence from these studies suggests that distress levels (intrusive thoughts about cancer, grief, anxiety and depression) may negatively impact how willing an individual is to follow screening protocols which are crucial for prevention and early detection (Codori et al., 2001; Jarvinen et al., 2000; Lerman et al., 1996). As well, there is some support for the fact that individuals with greater social supports and who live in families with open communication patterns are more likely to participate in regular screening (Johnson, Trimbath, Peterson, Griffin, & Giardiello, 2002; Keller et al., 2002).

Physicians and other primary health care providers also play a critical role in helping individuals follow recommended surveillance and screening protocols (Esplen et al., 2007; Hadley et al., 2004; Lindor et al., 2006; Stermer, Hodgson, Kavalier, Watts, & Jones, 2004). Clinicians should become actively involved in providing follow-up care to individuals after confirmation of HNPCC in the family. An important function is encouraging individuals to engage in recommended screening protocols (Lindor et al.; Lynch et al., 2008; Merg et al., 2005). Hadley et al. (2008) reported that HNPCC carriers screened more frequently when they share genetic testing results with family physicians.

Communication about HNPCC Risk

Hereditary cancers may affect family communication patterns as well as relationships among members. In many families, the affected person typically communicates information about hereditary cancers to other members. How accurate this

information is and how it is communicated have implications for how other family members understand their risk, become involved in genetic counseling and testing and decide to engage in screening (Gaff et al., 2005; Koehly et al., 2003; Peterson et al., 2003).

The literature suggests that individuals in HNPCC families may struggle with the responsibility of conferring information about a hereditary predisposition to cancer to other family members (Carlsson & Nilbert, 2007; Espen et al., 2007). Many individuals also experience concerns for family members, children or potential children (Bleiker et al., 2003; Carlsson & Nilbert; Claes et al., 2005; Espen et al.; Hadley et al., 2003). In arguing for more qualitative inquiries, McAllister (2002) noted that case studies and interview studies have identified complex family issues (i.e., pre-selection, scapegoating, survivor guilt, family communication difficulties and worry about children's risk) which may be difficult to detect with standardized questionnaires.

In a qualitative study Peterson et al. (2003) explored the psychosocial impact of predictive testing for HNPCC within the family context. These authors argued that the research base is limited on how families communicate information about genetic counseling and testing for HNPCC. The article describes how information is dispersed, when and under what circumstances information is shared, and how family members react to and act on the information received. Study findings indicated that communication about risk for HNPCC differed from that reported for other genetic diseases, like Huntington's disease. It was conjectured that cancer survivors are more willing to engage in open discussions and participate in such beneficial behaviors as counseling and testing. However, the authors noted that simply communicating information about risk potential

may not be enough and that follow-up and encouragement might be needed. In addition, family members with hereditary cancer who have contact with geneticists and genetic counselors may need to become intermediaries for at-risk relatives. As well, these individuals may need help with their advocacy role from spouses and unaffected family members. The authors also suggest that family members who become messengers may benefit from professional guidance.

Koehly et al. (2003) reported on the relational aspect of hereditary cancers from a psychosocial perspective. A study was designed to examine the broader family context and family systems in order to gain better insight into how these components influence family members' actions. The findings indicated a general tendency for individuals with a confirmed hereditary predisposition for cancer to discuss their experiences with genetic testing and counseling. As well, these communications tended to occur more frequently with first-degree relatives, close friends, and spouses. Finally, individuals from families with positive functional relationships (cohesion, positive leadership, communication, and active involvement) were more likely to engage in discussion about genetic counseling and genetic testing than those from families with lesser amounts of these characteristics.

Mesters et al. (2005) reported on the findings from a qualitative study that investigated individuals' experiences (reasons for, the process and extent of disclosure) with informing family members about HNPCC. Study findings suggested that individuals disclose information because of moral obligatory feelings and anticipated regret from not doing so, encouragement from medical care providers and/or significant others, and personal and family history of cancer (especially deaths of relatives). Participants discussed the hereditary information within the nuclear family only (first degree relatives

and spouses). The level of disclosure by study participants was limited to informing others about the condition's presence and the availability of genetic testing. More detailed information was left to the experts. Interestingly, the authors found that if the messenger encountered many negative responses, he or she was less likely to be willing to continue the disclosure process. However, it was also noted that if health care professionals stressed the importance of disclosure, this motivated the messengers.

Gaff et al. (2005) presented a brief overview of previous study findings on individuals' experiences with disclosure of information to family members. The authors noted that closeness of the family member genetically and socially, perceived relevancy for a family member, perceived potential for harm, age of the family member (children more likely to be informed), and gender of the informant (women more so than men) influence disclosure patterns. Other factors that may affect disclosure include beliefs about the importance of knowing and the right to know, as well as prior experience with others' reactions to distressful information.

The study by Gaff et al. (2005) was designed to assess the usefulness of a counseling session and information booklet in helping family members with hereditary cancer engage in full disclosure to all potentially at-risk relatives. Study findings indicated that male participants had a greater need for professional support than did females. In addition, despite not feeling the need for communication aids, participants supported having a letter summarizing the consultation results and an HNPCC information booklet to ensure that relatives received accurate information. Interestingly, although willing to disclose information about genetic testing, study participants did not

tell all at-risk family members. The authors noted that it did not seem to be a deliberate decision not to tell, but rather a result of poor communication pathways.

It is argued by some authors that individuals could benefit from ongoing contact with geneticists in order to receive up-to-date information about HNPCC and its management as well as support with disclosure to other family members (Collins et al., 2007; Gaff et al., 2005; Koehly et al., 2003; Mesters et al., 2005; Peterson et al., 2003). Given the wide range of potential implications for an individual who undergoes genetic testing for HNPCC, several authors advocate for a multidisciplinary counseling context in which to offer this service (Claes et al., 2005; Collins et al.; Meiser, 2005).

It is also recommended in the literature that genetic counselors should attempt to identify existing communication patterns within families and suggest ways that family members can take an active role in encouraging others to learn about their cancer risk and options for testing (Gaff et al., 2005; Koehly et al., 2003; Peterson et al., 2003). As genetic testing and counseling programs become more widely available, it is important to assess the psychological and social impact that this information has on individuals and families in order to provide the best professional support possible (Bleiker et al., 2003). Crucial in the process as well, is the autonomy of the individual and their ability to make informed decisions regarding testing and screening. Counselors must assess the needs and preferences of those they are counseling and adapt their sessions accordingly (Pieterse et al., 2005).

Summary

Research is needed to facilitate the development of sound theoretical models and provide the basis for appropriate genetic counseling. Otherwise, the provision of genetic testing may be ill-informed, ineffective, and counter-productive (Meiser, 2005). Future research should focus on ways to broaden the concept of the nuclear family in order to provide consistency in communication and disclosure, how written information provided by experts is being used, and helpful approaches or informant need for support. HNPCC-affected individuals and their relatives may benefit from specific guidance and advice on how to ensure that all those who are at risk receive the information they need, and that they understand the importance of receiving education and counseling to help manage their hereditary cancer risk (Peterson et al., 2003).

Instrument Development

Although genetic counseling strategies may be evidence-based, we know very little about what actually happens in counseling sessions especially in terms of their impact on individuals and families (Bleiker et al., 2003; Carlsson & Nilbert, 2007; Hadley et al., 2003; McAllister, 2001). The argument put forth by certain authors is that the existing research base on genetic testing decision-making is primarily descriptive and in need of theoretical models to guide inquiries into this area (Braithwaite et al., 2004; Etchegary, 2004; Gooding, Organista, Burack, & Biesecker, 2006; McAllister, 2001; Shiloh, 2006).

One group of researchers/theorists support the use of existing theory, especially social cognition models (Etchegary, 2004), Self-Regulatory Theory (Shiloh, 2006) and stress and coping models (Gooding et al., 2006). The position taken is that these models

will facilitate insight into the predictive power of key influencing and mediating factors in genetic testing uptake and quality cognitive, affective and behavioral outcomes post-genetic testing. In contrast, other authors identified the need for more theoretical and empirical research into factors influencing quality genetic testing decision-making and outcomes in the short- and long-term following confirmation of familial HNPCC (Bleiker et al., 2003; Marteau & Weinman, 2006; McAllister, 2001; Riper, 2005).

In addition, studies which have used standardized instruments for assessing psychological states (depression, impact of event, anxiety, worry and concerns) have not been consistent in their ability to predict psychosocial and behavioral outcomes from genetic testing (McAllister, 2002). In fact, the limited support for differences in distress levels between carriers and non-carriers of inherited diseases has been used to inform the practice of genetic counseling (McAllister, 2001). It is, therefore, important both theoretically and clinically to develop social psychological models that have greater explanatory power to inform the content of and the approaches used to deliver genetic services (McAllister, 2002).

According to Gregory and Way (2008) it is important to focus on the total illness experience – behavioral, social, psychological, and emotional – if we are to truly understand what it means to live with a chronic illness. In fact, greater interest in qualitative methods has emerged over the past decade in response to the increased recognition that is given to the role of psychosocial factors in shaping health outcomes (Gregory & Way). It is argued that the insights gleaned from qualitative data can be used to help researchers design instruments which are more sensitive to participants' meanings and interpretations (Coyle & Williams, 2000; Gregory & Way; McAllister, 2001). Gilgun

(2004) contends that solid qualitative research provides confidence in the content and face validity of clinical tools, and produces clinical tools that have excellent psychometric properties.

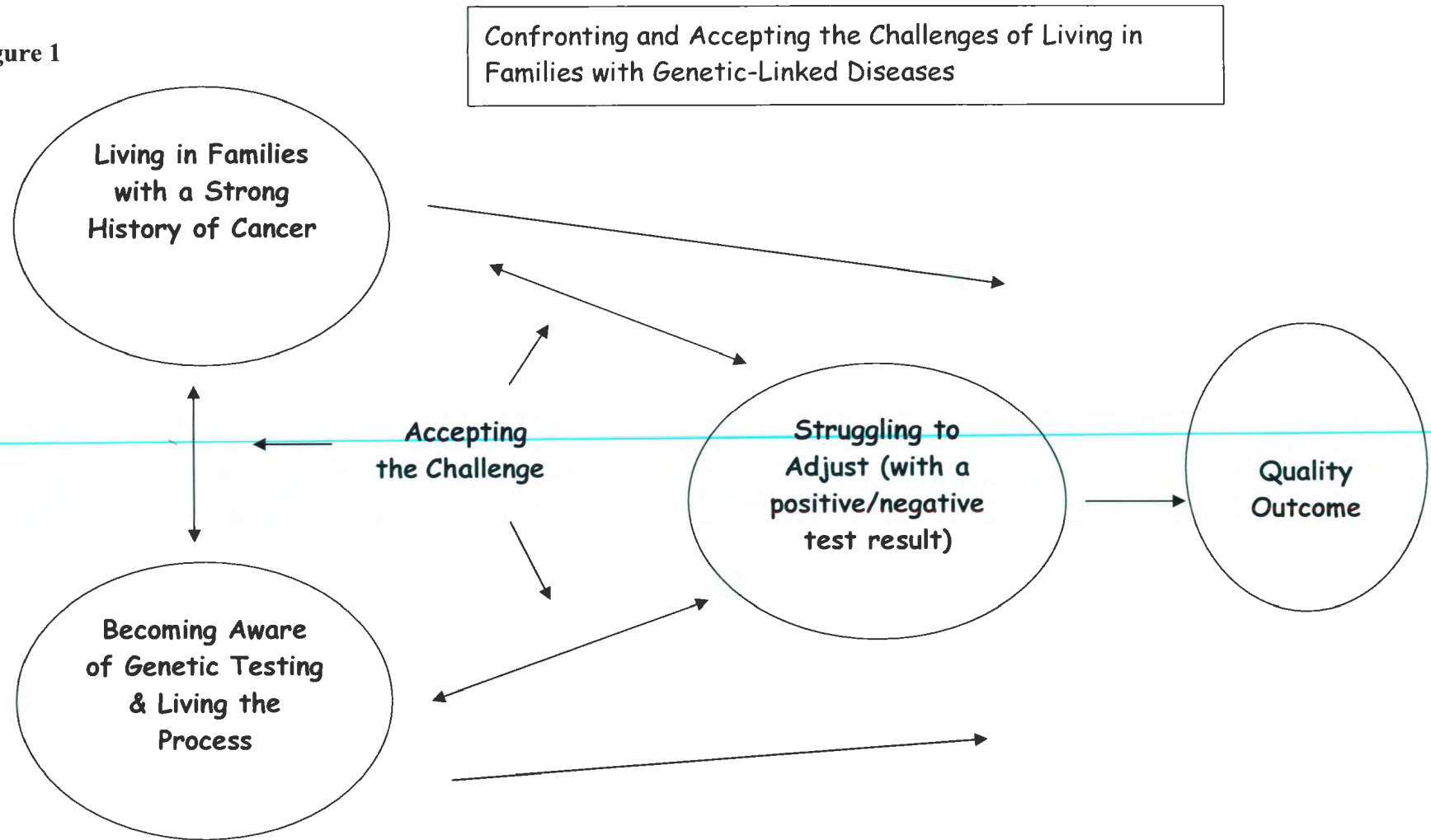
Based on the current review of literature, two researcher-developed instruments were identified that assessed motivation for genetic testing, as well as the concerns, needs and preferences of those seeking testing. Balmana et al. (2004) noted the presence of very few instruments which could be used to assess motivation and concerns related to genetic testing in hereditary cancer syndromes. These authors created an instrument based on their clinical experiences. Pieterse et al. (2005) also developed an instrument to measure needs and preferences in genetic counseling for hereditary cancer. This instrument, unlike the one by Balmana et al., was developed from the perspective of those seeking counseling.

Substantive Theory

Grounded theory methodology is useful for capturing basic psychosocial processes that form a substantive theory. The main objective of grounded theory is to facilitate a better understanding of human behavior and interactions within differing and similar contexts. The inductive – deductive approach is focused on generating theory as opposed to testing it (Gregory & Way, 2008; McAllister, 2001).

The framework used for the current study is presented in Figure 1. The figure depicts the substantive theory, *Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases*. It is based on the findings from a qualitative study conducted as part of the crCIHRt interdisciplinary project (Way et al., 2008).

Figure 1



The theory is comprised of three major constructs derived from the lived experiences of carriers and non-carriers of HNPCC: 1) living in families with a strong history of cancer, 2) becoming aware of genetic testing and living the process and, 3) struggling to adjust.

The substantive theory infers that the individual's family history and experience with cancer are important factors influencing how well individuals accept the hereditary link to cancer and are motivated to become involved in genetic testing. It is argued that this context is an important entry point marker because it influences the individual's cognitive thought processes in his or her search for meaning and understanding and, ultimately, impacts integration of the facts on emotional and behavioral levels.

The major underlying premise of the theory is that the first two constructs exert separate and interactive effects on each other and the third construct, struggling to adjust. All of the constructs are linked by the unifying thread of accepting the challenge. Psychosocial and behavioral adjustments waver in response to new challenges (e.g., adversities of screening protocols, progression to affected states, the suffering and early deaths of affected relatives). Adjustment is also influenced by ease of access to a supportive health care system (meaningful information, timely screening and treatment opportunities, psychosocial supports) and family/friendship networks. At the final step, it is inferred that living in families with a strong history of cancer and becoming aware of genetic testing and living the process exert a direct effect and indirect effect through struggling to adjust on quality outcome. As well, struggling to adjust exerts a direct effect on quality outcome.

The first construct, living in families with a strong history of cancer, gives a starting point for developing an appreciation of the intensity of the individuals' struggles with

multiple losses and how this shapes their search for meaning and understanding. It is comprised of two properties: 1) struggling with multiple losses – conflicting emotions and, 2) searching for meaning/understanding/certainty. These properties attempt to describe the experience of living with the presence of cancer in the family and the effect this has on family members. When individuals have struggled with the family cancer experiences, they have a sense of loss resulting from the worry and concern that goes far beyond the illness itself. The experiences of cancer in the family also influence the perception of risk for individuals.

The second construct, becoming aware of genetic testing and living the process, is a continuation of the genetic testing process to its end point – confirmed carrier or non-carrier status. It is comprised of four properties: 1) moving closer to puzzle completion, 2) meaning of genetic testing, 3) penetrance – dispelling beliefs and developing greater awareness, and 4) communicating with others – openness versus concealment.

Following contact with the geneticist, family members meaning base was expanded to include the increased probability of a hereditary component to the cancers. Although this provided them with greater certainty concerning the causal factor behind the cancers, uncertainty was also increased as they confronted a greater potential cancer risk for themselves and their families. The second property (meaning of genetic testing) focuses on how participants perceived the various ways in which they received their results, how they reacted to being informed about their carrier status, how well they understood their risk and how well they incorporated this into actionable knowledge in terms of screening. The third property deals with the processing of the information received regarding carrier status on both a cognitive and an emotional level. Existing beliefs about inheritance

contributed to acceptance or non-acceptance of these facts. The final property deals with how open individuals were about disclosing the results of genetic testing within and outside the family. There appears to be much variability regarding the ease of discussing this topic and the issue of who to tell and not to tell.

In this study, the research team used data from the first two constructs in the substantive theory to draft a scale. Living in families with a strong history of hereditary cancer, and becoming aware of genetic testing and living the process, were utilized in the development of the HD-GT scale. A more detailed description of the steps involved in the development and testing of this instrument are presented in Chapter 3.

Discussion of Literature

The literature reviewed for the current study highlighted the complex nature of what it means to live in a family with a known gene mutation for HNPCC and the many potential consequences that may emanate from this diagnosis. Based on this review, it is noteworthy that relatively few studies have examined the role that the family context plays in shaping perceptions of risk for self or others. In terms of interpreting personal risk and making decisions about obtaining genetic testing, research findings suggest that perceived risk for cancer may have more of an impact on genetic testing uptake than objective risk.

The research evidence suggests that many individuals with a family history of CRC experience high levels of perceived risk for developing the disease. This is associated with psychological distress prior to, during and following genetic testing. In addition,

delays experienced by some individuals while waiting for test results may enhance distress.

Study findings suggest that there may be a greater likelihood of genetic testing uptake in HNPCC families when there are personal experiences with cancer, a large number of affected relatives, and greater perceived risk for cancer. It is also conjectured that psychological engagement with the genetic testing process is believed to influence attitudes and motivation towards genetic testing for HNPCC. It has been conjectured that variations in the degree of psychological engagement with the genetic testing process may help explain differences in how individuals approach predictive DNA testing and react to their results.

The research evidence indicates that there are no adverse psychological effects on individuals as a result of engaging in predictive genetic testing. These findings have sometimes been attributed to the positive impact of genetic counseling. However, controversy exists about the definitiveness of this causal link. One confounding variable is the interactive effects of personal characteristics and genetic counseling information sessions on psychological outcomes. Other known confounders are age, gender, race, educational level, and the type and frequency of informal/formal social supports.

To date, the psychological impact of genetic testing has been more of a research focus than the behavioral impact. While the evidence is conclusive that regular screening is important for prevention and/or early detection of cancer, there is no guarantee that carriers will necessarily adopt appropriate screening behaviors following genetic testing. The few existing studies on behavioral tendencies post-genetic testing have found that most carriers follow recommended screening. Significantly, distress levels may

negatively impact adherence to recommended screening protocols, while greater social supports, the presence of open communication patterns in families, and the sharing of genetic testing results with family physicians are likely to positively impact screening behaviors.

Studies focusing on communication and disclosure patterns in HNPCC families have found that family dynamics seem to influence the target of communications and how much information is disclosed. Although there seems to be a greater tendency to disclose to those with closer ties to the individual, the information provided is often minimal. Interestingly, most individuals do not identify a need for support from health care providers to help them with disclosure.

Overall, the knowledge gaps surrounding the factors influencing genetic testing uptake and positive movement through the genetic testing process suggest the need for more theory to guide the content of and approaches taken during genetic counseling. Genetic counselors certainly need to be cognizant of how to relay information concerning HNPCC without increasing distress. As well, genetic counselors must take family communication patterns into consideration when planning their sessions.

Studies which have used standardized instruments to measure distress levels and perception of risk have not been consistent in their ability to predict psychosocial and behavioral outcomes from genetic testing, yet this literature has been used to inform the practice of genetic counseling. It is believed that the insight gleaned from qualitative data is more useful in designing instruments that will be able to detect the sensitive issues that emerge from living in families with a strong history of cancer. This knowledge can then

be used to develop social psychological models that will have greater explanatory power to inform the delivery of genetic services.

CHAPTER 3

Methodology

The pilot study was part of a larger study which is using a descriptive correlational design with longitudinal components to develop, validate and evaluate monitoring tools for individuals with genetic-based diseases. The primary focus of the current study was on generating data to assess the psychometric properties of the HD-GT scale. The content in this chapter is organized into two major sections. The first section provides a brief overview of the construction of the scale, content validation by the target population and experts, and scale readability. The second section highlights the steps taken to evaluate the psychometrics of the HD-GT scale.

Development of the HD-GT

Scale Development

Items for the HD-GT were derived from data matrices formulated from a qualitative data base generated from interviews with individuals (N = 39) comprising eight family groupings with a confirmed HNPCC presence. The HD-GT is designed to elicit data on two major constructs from the substantive theory, Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases, emerging from the data analysis (Way et al., 2008). The constructs of interest for the pilot study include: a) living in families with a strong history of cancer, b) and becoming aware of genetic testing and living the process.

The item generation phase was completed by a research team. This researcher was a member of the team. The process involved identifying potential item stems, reducing the number of item stems, reworking the item text and finalizing it, and selecting the best rating scale format to use with this population.

Initially, team members created a profile of frequency and priority ratings of properties and descriptors by individual participant and family groups. Data for this process were obtained from five family groupings. To provide inter-rater reliability, the principal investigator (PI) reviewed a sixth group in the same detail and validated the process. The remaining two groupings were subsequently reviewed for additional item stems. At the final stage, all items stems were combined to give a total of 267.

As item refinement proceeded, the emphasis was placed on conciseness and avoidance of negative wording, ambiguous terminology, jargon, value-laden words and double-barreled questions. Multiple drafts of items were reviewed and modified by the team to increase clarity and diminish redundancy. Following completion of this phase, a total of 68 items remained for potential inclusion in the HD-GT scale.

The next step involved identifying suitable Likert rating scales for testing the usefulness of the items. Initially, consideration was given to using multiple scales that focused on: a) frequency of occurrence of select events/conditions (never, rarely, sometimes, often, and almost always), b) the importance/difficulty/receptiveness of specified events/conditions (not at all, a little bit, moderately, quite a bit, extremely), and c) how satisfied/ concerned/confident/certain one was with events/situations (not at all, a little bit, moderately, quite a bit, extremely). The multiple selection options and anchors made item rating cumbersome. The decision was subsequently made to rework the items

to facilitate rating with one scale (not at all, a little bit, moderately, quite a bit, extremely).

Content Validation

Two types of experts were used for content validation. The HD-GT scale was first reviewed by two genetic counselors who work closely with individuals to prepare them for genetic testing and possible outcomes from this process. Genetic counselors were given a brief overview of the substantive theory, definitions and properties of the constructs and a copy of the scale. Input was first requested on the content relevancy (extremely, moderately, slightly, or irrelevant) of each item in terms of its ability to measure the properties of each construct. Secondly, they were asked to rate the effectiveness (very, moderately, poorly or not at all effective) of the 5-point Likert rating scale (i.e., not at all, a little bit, moderately, quite a bit, extremely) for allowing participants to indicate the importance/usefulness of items representing individual properties. Overall, the content experts confirmed item-relevancy for measuring appropriate constructs and the usefulness of the Likert rating scale. Recommendations included minor content changes to select items and eliminating three redundant items, leaving the final scale with 65 items for the next step.

The revised version of the HD-GT scale was subsequently content validated by two individuals (one carrier and one non-carrier of HNPCC) who had participated in the qualitative study. These two individuals commented on item clarity and relevancy, as well as the usefulness of the rating scale. Difficulties in scale administration, ambiguities in wording and the time required to complete the task were noted and addressed.

Scale Readability

Steps were taken by the research team to ensure that the HD-GT scale was at an appropriate reading level for the target population. Because this scale was developed to assess the experiences of individuals who have undergone predictive DNA testing for colorectal cancer, they have been exposed to terms such as HNPCC, carriers/non-carriers, inherited, generations, hereditary, genetic and geneticist/genetic counselor. These polysyllabic words and others are used frequently in the scale which does increase its final readability score.

Several methods were applied to assess the readability of the scale, including the Flesch-Kincaid Grade Level and Flesch Reading Ease, SMOG (Simple measure of gobbledook) and the Fog Index (Readability Formulas, n.d.). A grade less than 10 is recommended to ensure maximum reading ease and material comprehension. The findings indicated that the readability level of the HD-GT met the criteria.

Psychometric Evaluation of the HD-GT

Population and Sample

The target population was individuals at 50% risk for inheriting HNPCC who had participated in predictive testing and were informed of their carrier status. The goal was to recruit approximately equal numbers of carriers and non-carriers of the MSH2 mutation. Survey respondents were recruited from population-based probands comprising the Provincial Medical Genetics Program of Newfoundland and Labrador.

Three large pedigrees with MSH2 mutations on intron 5, exon 8 or exon 4 to 16 have been identified. Immediately prior to the pilot study, 272 carriers and 295 non-carriers had been confirmed from the Provincial Medical Genetics Program of Newfoundland and Labrador and entered into a Cancer Screening Data Base. This data base was developed for a component of the larger study which is retrospectively profiling the actual screening practices of carriers and non-carriers following genetic testing. The rationale for using this data base is that actual screening practices will be, ultimately, linked to the psychosocial and behavioral self-report data obtained over time following confirmation of the psychometric properties of the monitoring tools.

At the time of the pilot study, the information available on registrants comprising the Cancer Screening Data Base was reviewed to identify potential participants. Registrants excluded from consideration included those who did not have a confirmed carrier status (i.e., obligate carriers, presumed positive, or inconclusive results with unknown risk), had not participated in genetic testing, had died since their name was entered into the data base, had no contact information, or had refused to be contacted for research purposes. This left the research team with an initial list of 179 potential participants for the pilot study. Following additional inquiries, this number declined to 119 due to several factors (i.e., deceased, refused to review survey information to ascertain interest, family/personal distress, ill health, scheduled for surgery, living outside the province with no new contact information, or deemed to be mentally incompetent by a family member). Of the 119 individuals contacted by either this researcher or the research assistant and agreed to review the study materials, 75 (45 carriers and 30 non-

carriers) completed the survey at the time of the pilot study, resulting in a 63% response rate.

Procedure

Data collection commenced following receipt of ethical approval from the Human Investigation Committee, Faculty of Medicine, Memorial University as well as Eastern Health where the Provincial Medical Genetics Program of Newfoundland and Labrador is located (Appendix B). The pilot study phase spanned a three month time period from late February to late May 2008.

Telephone contact was initiated with potential respondents to inform them about the study and ascertain their willingness to receive additional information. The telephone script followed by the interviewers can be found in Appendix C. Individuals indicating an initial interest were forwarded packages consisting of a cover letter, a brief summary of the study, two consent forms (Appendix D) and the survey instrument (Appendix E).

Following receipt of the consent form, a follow-up telephone call was made by two members of the research team (this researcher and the research assistant) to determine the preferred mode of participation (face-to-face or telephone) and to schedule a mutually agreed upon time for survey completion. Some respondents returned the completed survey along with their consent forms.

The rationale for presenting respondents with optional modes for participating was based on the experience gained from implementing a previous quantitative study with this population (Way et al., 2008). The earlier study highlighted numerous problems using a self-administration approach to data collection, including a large amount of missing data

and inappropriate rating of certain items. This was true despite the fact that all of the scales used were designed for self-administration. Although face-to-face interviews are the preferred method to accommodate individual differences (illiteracy, diminished vision, or other problems which might interfere with a person's ability to complete a scale), total reliance on this approach is not practical in terms of time and cost given the large geographic dispersion of the target population (Polit & Beck, 2008). The pilot study was intended to provide additional data on the feasibility of using a mixed-methods approach with this population.

HD-GT Scale

The original HD-GT scale had ten sections with a total of 65 items. The content of this scale is based on two constructs – living in families with a strong history of cancer, and becoming aware of genetic testing and living the process. The first construct was to be assessed by the 17 items in sections A1 and A2 (Appendix E). Scale items in these two sections are intended to capture respondents' experiences with cancer and its treatment on a personal level and/or with family members, reactions to being informed about a possible MSH2 mutation within the family, and the impact on family relations.

The second construct was to be assessed with 48 items in sections A3 to A10 of the HD-GT (Appendix E). Scale items are intended to capture critical attitudes and feeling states at different junctures in the genetic testing process: a) lead-in period (motivation to participate, cognitive and emotional preparedness), b) during (desire to know test results, emotional difficulties while waiting, concerns over others reluctance to participate), and c) post-test period (adequacy of supports while receiving results, understanding of risks,

awareness of family cancer patterns, personal/family communications/disclosure around HNPCC risk).

Ethical Considerations

Several steps were taken to protect participant rights in the current study. All individuals meeting the inclusion criteria were contacted by either this researcher or the research assistant. Individuals who agreed to participate were given three opportunities to consider their agreement to participate in the project. First, all individuals were asked in the initial telephone call if they would like to participate. Those who agreed were then sent study materials (Appendix D & E) and were asked to complete the enclosed consent and forward it to the research team in the return envelope provided. Returned consent forms were perused for appropriate completion and then follow-up telephone contact initiated. Before proceeding to interview scheduling, the interviewer determined that potential respondents understood what they had consented to do and were still willing to proceed with the study. This approach ensured that potential respondents were not subjected to coercion or undue influence.

Confidentiality of all data has been maintained by assigning a numerical code to all forms and instruments. A master list of names and corresponding codes is being kept in a locked filing cabinet accessible only to the research team.

Data Analysis

Data were coded and entered into the Statistical Package for the Social Sciences (SPSS) for analysis. Descriptive statistics were used to create a profile of respondents'

scores on all study instruments. Exploratory factor analysis was used to assess subscale structure. The initial factor solution and varimax rotation failed to generate factors that aligned with the proposed constructs of the substantive theory in a meaningful way. This could be due, in part, to the small sample size.

As a result of the non-usefulness of the factor analysis procedure, a correlation matrix was generated of all HD-GT items to search for item groupings as recommended by Ware and Gandek (1998). Projected subscales were then constructed and their items evaluated for adherence to Likert scaling assumptions. At the first step, the assumption concerning the appropriateness of using particular items to create a summative score (approximate equivalence of means and variances, use of all response choices in the rating scale, amount of missing data, and approximate symmetry in response distribution) were assessed. At the second step, a multi-item/multi-trait correlation matrix was generated to assess three additional assumptions (linearity, item-convergent validity and item-discriminant validity). At the final step, subscale scores were assessed in terms of ceiling and floor effects, approximate symmetry, internal consistency and inter-correlations.

CHAPTER 4

Results

Study findings are presented in three sections. The first section presents a summary of the descriptive findings on the sample and HD-GT scale. The second section presents findings on the reliability and validity of the HD-GT scale. The final section presents a brief discussion on the study findings.

Descriptive Profile

Study findings are organized under six major subsections. First, a brief overview is presented on the personal characteristics (demographics and illness-related) of the sample. Second, the outcome of variant scale administration is presented. Third, the results are presented on the final subscale structure derived from inter-item correlation analyses. Fourth, a detailed summary is presented of the data quality and how well individual items met scaling assumptions. The fifth subsection focuses on the subscales of the HD-GT and examines hypothesized item groupings in terms of internal consistency, equality of item-scale correlations, and item discriminant validity. The final subsection presents the findings on the subscale scores for the HD-GT.

Personal Characteristics

The sample for the pilot was comprised of 75 respondents. The mean age was 52.82 (SD = 13.69), with a range from 24 to 83 years. The majority of respondents were female (66.67%).

Table 1 summarizes illness-related characteristics of the sample. Most participants were carriers of the HNPCC gene (60%) but unaffected by cancer at the time of the study (55.56%). The dominant mutation type was the intron 5 splice site mutation (72%). Of those respondents who reported having had a bout of cancer ($n = 21$), the most predominant type was colon (61.90%) either alone or in combination with other cancers. The vast majority of respondents (93.33%) were actively involved in cancer screening.

HD-GT Scale Administration

The HD-GT was administered to participants in one of three ways: face-to-face, telephone or self-administered. Data completeness was similar for all three methods, indicating that it is possible to administer this scale under variant conditions.

Subscale Structure

Following exploratory factor analysis, all of the items for the conjectured theoretical factors were not lining up as expected. The decision was then made to generate a correlation matrix for all of the HD-GT scale items. Each item was subsequently analyzed for the strength and significance of its correlation with all other items.

A table summary was constructed of items correlating with other items within the set cutoff ranges (i.e., $>.40$ and $.30$ to $.40$). This exercise was the primary basis for final selection of items for each subscale. The inclusion of other items not meeting these criteria was based on conceptual logic around item content for designated subscales based on the substantive theory.

Table 1. Illness-Related Characteristics (N = 75)

	n	%
Carrier Status		
Carrier	45	60.00
Cancer Presence – Yes	20	44.44
Cancer Presence – No	25	55.56
Non-carrier	30	40.00
Cancer Presence – Yes	1	03.33
Cancer Presence – No	29	96.67
Mutation Type		
Intron 5 Splice Site	54	72.00
Exon 8 Deletion	21	28.00
Cancer Type		
Colon Only	5	23.81
Reproductive Only	5	23.81
Colon & Reproductive	3	14.29
Colon & Other	5	23.81
Other Only	3	14.29
Cancer Screening		
Yes	70	93.33
No	5	06.67

Following completion of this stage of the analysis, 13 items failed to correlate in a consistent manner with other items in terms of the set cut-off range. These items were subsequently removed, leaving the final version of the HD-GT scale with nine subscales comprised of 52 items. The subscales for the HD-GT and their item totals are summarized in Table 2.

Data Quality and Item-level Summated Scale Assumptions

Data Quality. A useful measure of data quality is the amount of missing data for each item and projected subscales of a summated rating scale (Ware & Gandek, 1998). Another component of data quality addresses the frequency distribution or spread of scores across the steps (response choices) of a rating scale (Ware & Gandek). This information is examined for two reasons: 1) usefulness of all steps in a rating scale and, 2) variability and symmetry of the data.

Table 3 summarizes the findings for the 52 items and nine subscales of the HD-GT. Missing data for individual items were random and minimal, ranging from 0% to 4%. Although there is no consensus on what constitutes extensive missing data (from 10% – 40%) on any given item or variable, it is generally agreed that what is more important is whether the pattern is systematic or random in nature (Fox-Wasylyshyn & El-Masri, 2005).

As well, the majority of respondents (74.67%) had complete data for all of the subscales of the HD-GT. The percent of respondents with complete data for individual

Table 2. Theoretical Constructs and HD-GT Subscales

Theoretical Constructs	HD-GT Subscales
Construct 1 - Living in Families	Impact of Familial Cancer (IFC) – 9 items
Construct 2 – Genetic Testing	Family Challenges Genetic Testing (FCGT) – 6 items Genetic Testing Preparation (GTP) – 9 items Wait Time Concerns (WC) – 6 items Support for Genetic Testing Results (SGTR) – 5 items Understanding Risk (UR) – 5 items Transmission Beliefs (TB) – 4 items Communications around Genetic Link (CGL) – 4 items Disclosure Issues (DI) – 4 items

Table 3. HD-GT Data Quality and Item-level Descriptive Statistics

Scale Items	X	SD	Missing (%)	Response Values Frequency				
				0	1	2	3	4
Impact of Familial Cancer (IFC)								
• Cancers more frequent & at younger age (IFC11_R)	1.25	1.36	4.0	28	20	11	4	9
• Memories of cancer suffering (IFC12_R)	1.03	1.25	2.7	35	16	12	5	5
• Difficulty accepting cancer motivate to know (IFC13_R)	1.37	1.35	2.7	27	14	18	6	8
• Scary observing same cancer pattern (IFC14_R)	1.01	1.33	2.7	38	15	7	7	6
• Draining to lose members to cancer (IFC15_R)	1.92	1.50	2.7	18	13	16	9	17
• Cancer suffering/death worried about self (IFC16_R)	1.75	1.45	2.7	21	10	22	6	14
• Worry re multiple cancers in one member (IFC17_R)	1.58	1.42	2.7	21	20	13	7	12
• Stress of cancer altered family relations (IFC18_R)	2.53	1.42	2.7	7	14	13	11	28
• Screening reminder of personal risk (IFC26_R)	1.86	1.47	2.7	20	9	20	9	15
Family Challenges Genetic Testing (FCGT)								
• Genetic testing [GT] important for everyone (FCGT41)	3.68	0.85	1.3	2	1	3	7	61
• Concerned about those who refuse GT (FCGT42)	3.09	1.21	1.3	4	6	8	17	39
• Concern understand risk when refuse GT (FCGT43)	3.12	1.20	2.7	4	5	9	15	40
• Refuse GT – fearful of knowing status (FCGT44)	3.34	1.08	2.7	4	2	4	18	45
• Important everyone informed of HNPCC (FCGT105)	3.85	0.56	0.0	1	0	1	5	68
• Better knowing status than not knowing (FCGT76)	3.73	0.84	0.0	2	2	1	4	66
Genetic Testing Preparation (GTP)								
• Timely GT info from genetic personnel (GTP31)	3.36	1.06	0.0	2	4	9	10	50
• Adequate info about GT process (GTP32)	3.39	0.99	0.0	2	3	7	15	48
• Important to know one’s HNPCC status (GTP34)	3.36	1.08	1.3	2	5	7	10	50
• Understand HNPCC risk and accept GT (GTP35)	3.58	0.74	1.3	0	1	8	12	53

• Help offspring to know HNPCC status (GTP36)	3.62	0.87	1.3	2	1	4	9	58
• Need genetic personnel support during GT (GTP37)	3.07	1.28	2.7	6	5	6	17	39
• Need support from family/friends (GTP38)	3.37	0.99	0.0	3	1	7	18	46
• Relieved study investigating familial mutation (GTP23)	3.14	1.12	4.0	2	5	13	13	39
• Positive attitude toward GT (GTP24)	3.55	0.86	1.3	0	5	3	12	54
Wait-Time Concerns (WC)								
• Trying time thinking about HNPCC status (WC51_R)	2.03	1.48	1.3	17	13	10	19	15
• Not prepared for long wait for GT results (WC52_R)	2.65	1.43	0.0	8	12	9	15	31
• Uncertain about medium for results receipt (WC53_R)	3.07	1.32	0.0	5	7	11	7	45
• Dwelling on reaction to GT results (WC54_R)	2.43	1.39	0.0	8	15	12	17	23
• Question ability to understand GT results (WC55_R)	2.59	1.44	0.0	9	11	12	13	30
• Geneticist contact increased perceived risk (WC22_R)	1.70	1.55	2.7	23	15	13	5	17
Support for Genetic Testing Results (SGTR)								
• Family/friend present (SGTR61)	2.32	1.56	0.0	16	9	10	15	25
• Call from genetic personnel prior to receipt (SGTR62)	2.01	1.57	1.3	21	9	10	16	18
• Face to face contact to receive results (SGTR63)	2.53	1.49	0.0	12	7	15	11	30
• Receive letter explaining HNPCC status (SGTR64)	3.28	1.17	0.0	5	3	4	17	46
• Follow-up important re lifestyle/screening (SGTR75)	2.41	1.51	1.3	14	8	10	18	24
Understanding Risk (UR)								
• Regular screening for cancer detection (UR82)	3.83	0.50	4.0	0	1	1	7	63
• Appropriate screening timely cancer detection (UR83)	3.89	0.36	2.7	0	0	1	6	66
• Early detection improve disease management (UR84)	3.88	0.37	2.7	0	0	1	7	65
• Healthy living/positive attitude improve QOL (UR85)	3.55	0.86	1.3	2	0	6	13	53
• Self-responsible for healthy living/screening (UR86)	3.82	0.59	2.7	1	0	1	7	64

Transmission Belief (TB)								
• Younger age first HNPCC cancer (TB91)	3.01	1.27	2.7	7	3	7	21	35
• Men/women different type first cancer (TB92)	2.37	1.34	2.7	12	4	18	23	16
• Different types cancer today than before (TB93)	2.45	1.38	1.3	11	6	17	19	21
• Greater numbers of family members with cancer than before (TB94)	2.38	1.32	1.3	10	7	20	19	18
Communication around Genetic Link (CGL)								
• Young people open to HNPCC info (CGL103)	2.84	1.12	1.3	5	3	14	29	23
• Young people understand own HNPCC risk (CGL104)	2.74	1.15	2.7	5	5	15	27	21
• All family open to HNPCC info (CGL107)	3.08	1.03	1.3	1	4	18	16	35
• All family understand own HNPCC risk (CGL108)	3.11	0.95	1.3	1	3	15	24	32
Disclosure Issues (DI)								
• Difficult inform young people about HNPCC risk (DI102)	2.67	1.41	0.0	9	9	9	19	29
• Difficult inform family members about HNPCC risk (DI106)	2.23	1.57	1.3	19	5	11	18	21
• Genetic personnel guidance needed to inform (DI109)	2.78	1.40	1.3	8	8	9	16	33
• Protect rights of others relating HNPCC risk (DI110)	3.35	1.07	0.0	4	0	10	13	48

Note: 1) Response choice values are: 0 = Not at all; 1 = A little bit; 2 = Moderately; 3 = Quite a bit; 4 = Extremely.

2) Items with the designate _R are reverse scored.

subscales ranged from 89.33% for the GTP to 97.33% for the SGTR and DI (data not shown). The minimum and random amount of missing data for this pilot study suggests that overall the scale items were not difficult to understand or interpret (Ware & Gandek, 1998).

The findings also suggest that there were minimal issues with interpreting the response choices. All response choices were used for most items (82.70%). The data also depict wide variability across the rating scale and approximate a symmetrical distribution. The subscale items with minimal to no use of certain response choices were expected. That is, most individuals are expected to attach high importance to being adequately prepared for genetic testing (GTP), to understand their cancer risk (UR), to belong to families that openly communicate about the genetic disease risk (CGL), and to experience few difficulties informing other members about their potential risk (DI).

Item-level scaling assumptions. This section examines the patterns observed with item means and standard deviations for each hypothesized subscale of the HD-GT. Items means and standard deviations within each subscale are approximately equivalent (Table 3). There are important exceptions however which require further elaboration. In the IFC subscale, items 15, 16, 18 and 26 have higher mean scores and greater variance than the remaining items. This finding is expected given that these items are more focused on assessing feeling states and perceived implications for the self.

The higher mean scores and lower variances observed for items 41, 105 and 76 comprising the FCGT subscale were also expected since their content focuses on the importance of genetic testing for everyone presumed to be at high risk for cancer. As well, the lower mean scores for items 37 and 62 in the GTP and SGTR subscales,

respectively, are expected because not everyone needs ongoing support from the geneticist/genetic counselor throughout the genetic testing process or expects a call prior to receiving their results. Finally, the higher mean scores for items 53, 64, 91 and 110 from the WC, SGTR, TB and DI subscales, respectively, were also expected. That is, it is normal to experience uncertainty about the medium to be used for conveying results (WC) and everyone expects to receive a letter detailing the information conveyed regarding their results (SGTR). As well, the evidence in local, national and international families indicate that the age for first onset of HNPCC is declining (TB) and there are more concerns today about protecting the rights of others (DI) (Lynch & de la Chapelle, 2003; Lynch et al., 2008; Merg et al., 2005; Stuckless et al., 2007).

Scale Level Assumptions

Following construction of the proposed scales, a second correlation matrix was generated of all items and projected subscales. In addition to this multi-trait/multi-item correlation matrix, internal consistency was generated for each subscale to derive the corrected item-total correlations. All of these steps were necessary to determine the best subscale for individual items. Table 4 contains a summary of these findings which provide support for the Likert scaling assumptions (i.e., item internal consistency, equality of item-scale correlations and item discriminant validity) as discussed in the following sections.

Table 4. Correlation Matrix of HD-GT Items with each Subscale

Scale	X	SD	IFC	FCGT	Pearson Item Scale Correlation (*Corrected)							
					GTP	WC	SGTR	UR	TB	CGL	DI	
Impact of Familial Cancer (IFC)												
• IFC11_R	1.25	1.36	0.64*	0.10	0.27	0.21	0.20	0.22	0.38	0.37	0.13	
• IFC12_R	1.05	1.25	0.54*	0.30	0.15	0.16	0.17	0.00	0.21	0.47	0.13	
• IFC13_R	1.37	1.35	0.71*	0.30	0.25	0.29	0.34	0.15	0.42	0.41	0.24	
• IFC14_R	1.01	1.33	0.80*	0.31	0.34	0.31	0.30	0.23	0.46	0.46	0.17	
• IFC15_R	1.92	1.50	0.70*	0.20	0.24	0.35	0.34	0.06	0.50	0.39	0.22	
• IFC16_R	1.75	1.45	0.75*	0.30	0.31	0.40	0.35	0.17	0.48	0.43	0.24	
• IFC17_R	1.58	1.42	0.69*	0.24	0.32	0.20	0.23	0.23	0.31	0.60	0.01	
• IFC18_R	2.53	1.42	0.51*	0.24	0.27	0.48	0.28	0.04	0.47	0.36	0.33	
• IFC26_R	1.86	1.48	0.51*	0.38	0.35	0.43	0.32	0.14	0.43	0.37	0.17	
Family Challenges Genetic Testing (FCGT)												
• FCGT 41	3.68	0.85	0.32	0.67*	0.72	0.31	0.51	0.47	0.15	0.28	0.25	
• FCGT 42	3.09	1.21	0.39	0.67*	0.50	0.48	0.52	0.42	0.37	0.25	0.28	
• FCGT 43	3.12	1.20	0.21	0.64*	0.41	0.37	0.26	0.30	0.21	0.23	0.17	
• FCGT 44	3.34	1.08	0.29	0.61*	0.37	0.46	0.47	0.34	0.24	0.46	0.31	
• FCGT 76	3.73	0.84	0.16	0.44*	0.10	0.27	0.33	0.03	0.21	0.28	0.00	
• FCGT 105	3.85	0.56	0.44	0.53*	0.61	0.27	0.40	0.44	0.23	0.39	0.25	

Scale	Pearson Item Scale Correlation (*Corrected)											
	X	SD	IFC	FCGT	GTP	WC	SGTR	UR	TB	CGL	DI	
Genetic Testing Preparation (GTP)												
• GTP31	3.36	1.06	0.36	0.38	0.87*	0.41	0.50	0.45	0.14	0.26	0.39	
• GTP32	3.39	0.99	0.39	0.37	0.82*	0.45	0.53	0.41	0.20	0.32	0.29	
• GTP34	3.36	1.08	0.22	0.47	0.81*	0.29	0.38	0.42	0.04	0.23	0.15	
• GTP35	3.58	0.74	0.34	0.45	0.88*	0.39	0.46	0.49	0.09	0.25	0.19	
• GTP36	3.62	0.87	0.30	0.36	0.74*	0.23	0.41	0.31	0.03	0.08	0.36	
• GTP37	3.07	1.28	0.30	0.32	0.67*	0.44	0.69	0.41	0.15	0.19	0.46	
• GTP38	3.37	0.99	0.45	0.38	0.71*	0.43	0.58	0.52	0.15	0.41	0.39	
• GTP23	3.14	1.12	0.29	0.44	0.60*	0.28	0.42	0.40	0.04	0.32	0.13	
• GTP24	3.55	0.86	0.10	0.35	0.32*	0.15	0.24	0.23	0.13	0.12	0.01	
Wait-Time Concerns (WC)												
• WC51_R	2.03	1.48	0.44	0.41	0.44	0.77*	0.61	0.31	0.40	0.34	0.53	
• WC52_R	2.65	1.43	0.33	0.34	0.32	0.66*	0.36	0.23	0.43	0.28	0.47	
• WC53_R	3.07	1.32	0.21	0.32	0.21	0.52*	0.23	0.21	0.36	0.16	0.39	
• WC54_R	2.43	1.39	0.39	0.33	0.44	0.76*	0.57	0.27	0.37	0.26	0.55	
• WC55_R	2.59	1.44	0.28	0.45	0.37	0.67*	0.45	0.29	0.37	0.32	0.36	
• WC22_R	1.70	1.55	0.41	0.43	0.29	0.54*	0.24	0.15	0.27	0.40	0.24	

Scale	Pearson Item Scale Correlation (*Corrected)										
	X	SD	IFC	FCGT	GTP	WC	SGTR	UR	TB	CGL	DI
Support for Genetic Testing Results (SGTR)											
• SGTR61	2.32	1.56	0.29	0.34	0.42	0.45	0.70*	0.26	0.16	0.23	0.46
• SGTR62	2.01	1.57	0.40	0.31	0.47	0.45	0.66*	0.38	0.33	0.32	0.52
• SGTR63	2.53	1.49	0.26	0.31	0.48	0.46	0.69*	0.19	0.22	0.19	0.35
• SGTR64	3.28	1.17	0.42	0.39	0.52	0.34	0.55*	0.41	0.35	0.40	0.27
• SGTR75	2.40	1.50	0.26	0.41	0.43	0.34	0.56*	0.24	0.31	0.14	0.27
Understanding Risk (UR)											
• UR82	3.83	0.50	0.14	0.02	0.28	0.18	0.11	0.56*	0.12	0.23	0.04
• UR83	3.89	0.36	0.11	0.06	0.31	0.14	0.16	0.62*	0.12	0.17	0.02
• UR84	3.87	0.37	0.05	0.16	0.15	0.22	0.15	0.60*	0.17	0.17	0.04
• UR85	3.55	0.86	0.25	0.51	0.54	0.33	0.44	0.58*	0.09	0.17	0.25
• UR86	3.82	0.59	0.31	0.45	0.68	0.22	0.38	0.59*	0.03	0.25	0.22

Scale	Pearson Item Scale Correlation (*Corrected)										
	X	SD	IFC	FCGT	GTP	WC	SGTR	UR	TB	CGL	DI
Transmission Belief (TB)											
• TB91	3.01	1.27	0.42	0.29	0.01	0.29	0.26	0.00	0.65*	0.28	0.16
• TB92	2.37	1.34	0.38	0.20	0.12	0.26	0.26	0.01	0.68*	0.15	0.11
• TB93	2.45	1.37	0.34	0.27	0.15	0.41	0.26	0.26	0.62*	0.31	0.26
• TB94	2.38	1.32	0.48	0.31	0.07	0.43	0.19	0.17	0.58*	0.35	0.26
Communication around Genetic Link (CGL)											
• CGL103	2.83	1.12	0.52	0.35	0.33	0.31	0.27	0.20	0.35	0.64*	0.08
• CGL104	2.74	1.16	0.52	0.39	0.35	0.35	0.22	0.22	0.35	0.81*	0.13
• CGL107	3.08	1.03	0.35	0.44	0.14	0.27	0.27	0.18	0.24	0.67*	0.06
• CGL108	3.11	0.95	0.42	0.29	0.20	0.32	0.30	0.24	0.27	0.77*	0.12
Disclosure Issues (DI)											
• DI102	2.67	1.40	0.25	0.21	0.30	0.43	0.37	0.12	0.19	0.20	0.66*
• DI106	2.23	1.57	0.08	0.12	0.16	0.44	0.27	0.11	0.20	0.09	0.64*
• DI109	2.78	1.40	0.28	0.33	0.42	0.49	0.59	0.22	0.26	0.14	0.57*
• DI110	3.34	1.07	0.14	0.17	0.06	0.31	0.24	0.06	0.19	0.10	0.47*

Note: IFC = Impact of Familial Cancer; FCGT = Family Challenges Genetic Testing; GTP = Genetic Testing Preparation; WC = Wait-Time Concerns; SGTR = Support for Genetic Testing Results; UR = Understanding Risk; TB = Transmission Beliefs; CGL = Communication around Genetic Link; DI = Disclosure Issues.

Item internal consistency. Item internal consistency assesses the degree to which an item correlates with its scale score in a substantive, linear fashion. Such correlations are derived by excluding relevant items from their intended scales, calculating scale scores from remaining items, and then correlating removed items with the new scale scores. This corrected item-total correlation reduces inflation levels and can be generated in SPSS by the Cronbach's alpha procedure for reliability. Corrected item-total correlations for individual items and their subscales are highlighted by an asterisk in Table 4.

As shown in Table 4, the corrected correlation of items with their respective scales was greater than .40 for the most part. The only exception was item 24 of the GTP subscale which fell within the .30 and .40 range. This item measures motivation to become involved in genetic testing prior to actually doing so and is therefore an important factor in the entire process.

Overall the findings support the presence of substantial correlations between HD-GT items and their hypothesized scales. Thus, the data fulfill the criteria for the item internal consistency assumption.

Equality of item-scale correlations. This assumption addresses the proximity of values for all of the item-scale correlations within a hypothesized scale. The best scale contains item-scale correlations that are roughly equal and ideally fall within the .40 to .70 range (Ware & Gandek, 1998). The reader is again referred to the corrected item-total correlations for individual items and their subscales in the columns with asterisks in Table 4.

For the majority of HD-GT subscales, the corrected-item total correlations fall within an acceptable range. There are some exceptions however. The items that appear to

be contributing more to their various scales than other items include items 14 and 16 of the IFC subscale, items 51 and 54 of the WC subscale, and items 104 and 108 from the CGL. It is interesting that these items are dealing with emotional content which may be responsible for the observed discrepancies. The other subscale evidencing variable contributions of items to the scale score is the GTP. This finding is expected to a degree since item content is focused on different aspects of the genetic testing process (i.e., importance of adequate and timely information versus importance of knowing and understanding versus the importance of having adequate supports and being emotionally prepared for genetic testing).

It is important to note that correlations are only one factor to consider when placing items in a particular scale. There are also content implications of particular items which must be considered when decisions are made about their retention or exclusion from a scale. Finally, current findings are based on data obtained from a small sample.

Item discriminant validity. This assumption takes the logic of equality of item-scale correlations (magnitude of correlation with hypothesized scale) a step further to examine the strength of item correlations with other scales in a matrix that it is not intended to measure. In this case, the objective is for each item to have a stronger correlation with its hypothesized scale than with related scales comprising a matrix. Study findings for the HD-GT are summarized in Table 4.

For the most part, the HD-GT items evidenced good discriminatory power across the subscales. The exceptions were items 105 and 41 of the FCGT subscale, items 24 and 37 of the GTP subscale, item 86 of the UR subscale and item 109 of the DI subscale.

Scale Level Descriptive Statistics

Total subscale scores were constructed for each participant following confirmation of item scaling assumptions. Consideration was first given to the impact of select sample characteristics on subscale scores. At the second step, the properties of the subscales were examined with special attention given to the logic of mean and standard deviation scores. Table 5 summarizes the subscale score for carriers and non-carriers but not the total sample.

Comparability of scale scores. It was hypothesized that subscale means should be approximately equal within the sample based on demographic and illness-related characteristics. The reader is reminded that two of the subscales, IFC and WC, are reversed scored. The t-test of difference and correlation tests assessed the impact of select factors on subscale scores. No significant effect was detected for exon type, cancer presence, gender or age ($p > .05$) (data not shown).

The IFC, FCGT and CGL subscales evidenced significant differences for HNPCC status ($p < .05$). Table 5 summarizes these findings. Specifically, carriers more so than non-carriers reported less impact of family cancer on genetic testing decision-making (IFC) but attached greater importance to having at-risk family members participate in genetic testing (FCGT). As well, carriers perceived family members to be less open to and accepting of information about HNPCC than non-carriers.

Scale properties. Subscale means, standard deviations, lowest and highest scores and score ranges were examined for both raw and transformed scores. The focus here was on the logic behind the distribution of subscale scores.

Table 5. HD-GT Subscale Scores by HNPCC Status

Subscales	Range	Carriers M (SD)	Non-Carriers M (SD)	t
Impact of Familial Cancer (IFC)	0-36	16.37 (9.75)	11.71 (7.66)	2.13 (p=.036)
Family Challenges Genetic Testing (FCGT)	0-28	19.95 (4.87)	22.51 (1.99)	-3.09 (p=.003)
Genetic Testing Preparation (GTP)	0-36	30.08 (7.30)	31.46 (6.15)	-0.82 (p=.416)
Wait time Concerns (WC)	0-24	15.57 (6.6)	12.67 (6.49)	1.85 (p=.068)
Support for Genetic Testing Results (SGTR)	0-20	12.14 (6.21)	13.10 (4.75)	-0.71 (p=.479)
Understanding Risk (UR)	0-20	18.80 (2.25)	19.25 (1.71)	-0.91 (p=.364)
Transmission Beliefs (TB)	0-16	9.77 (3.86)	10.64 (3.84)	-0.93 (p=.354)
Communication around Genetic Link (CGL)	0-16	11.0 (3.83)	13.19 (2.72)	-2.82 (p=.006)
Disclosure Issues (DI)	0-16	10.58 (4.34)	11.68 (4.12)	-1.08 (p=.286)

For most subscales, higher scores are reflective of greater importance, understanding, awareness, and openness and acceptance. The exceptions are the reverse scored IFC and WC where higher scores reflect less impact of familial cancer and less wait-time emotional challenges, respectively.

The pattern of mean scores and standard deviations for each subscale is summarized in Table 6. The low to moderate mean score for IFC indicates that familial cancer history had a significant impact on genetic testing decision-making. The high mean scores for FCGT and GTP suggest that great importance was attached to having at-risk family members participate in genetic testing and to be adequately informed and emotionally prepared for this process. The moderate mean score for WC indicates that a fair number of respondents were challenged emotionally while waiting for genetic testing results. Comparatively, the moderate mean score for SGTR suggests that not everyone attaches high importance to having family and/or health care providers present during receipt of genetic testing results. The UR has a relatively high mean score which is significant since this suggests that most respondents understood the importance of regular screening and healthy lifestyles. The three remaining scales – TB, CGL and DI – have mean scores that fall within the moderate to high range. These findings suggest that one-third to one-quarter of respondents did not have a complete picture of cancer profiles in the family or perceive that family members of all ages were open to and accepting of HNPCC information, and encountered some difficulties around disclosure.

Table 6. Descriptive Statistics for Raw and Transformed Subscale Scores of the HD-GT Scale

Raw Scores

Subscale	Mean	SD	Observed/Possible Values			Floor (%)	Ceiling (%)
			Lowest	Highest	Range		
Impact of Familial Cancer (IFC)	14.53	9.21	0/0	36/36	36/36	1.4	2.8
Family Challenges Genetic Testing (FCGT)	20.99	4.15	4/0	24/24	20/28	1.4	36.1
Genetic Testing Preparation (GTP)	30.66	6.83	5/0	36/36	31/36	1.5	29.9
Wait-Time Concerns (WC)	14.36	6.67	0/0	24/24	24/24	1.4	6.9
Support for Genetic Testing Results (SGTR)	12.52	5.66	0/0	20/20	20/20	4.1	11.0
Understanding Risk (UR)	18.97	2.06	11/0	20/20	9/20	1.4	70.8
Transmission Beliefs (TB)	9.73	3.99	0/0	16/16	16/16	2.8	6.9
Communications around Genetic Link (CGL)	11.82	3.60	0/0	16/16	16/16	1.4	15.3
Disclosure Issues (DI)	11.00	4.26	0/0	16/16	16/16	1.4	17.8

Transformed Scores (0-100)

Subscale	Mean	SD	Observed/Possible Values			Floor (%)	Ceiling (%)
			Lowest	Highest	Range		
IFC	40.38	25.59	0/0	100/100	100.00/100	1.4	2.8
FCGT	87.44	17.29	16.67/0	100/100	83.33/100	1.4	36.1
GTP	85.16	18.96	13.89/0	100/100	86.11/100	1.5	29.9
WC	59.84	27.79	0/0	100/100	100.00/100	1.4	6.9
SGTR	62.60	28.30	0/0	100/100	100.00/100	4.1	11.0
UR	94.86	10.28	55.00/0	100/100	45.00/100	1.4	70.8
TB	63.19	24.07	0/0	100/100	100.00/100	2.8	6.9
CGL	73.87	22.48	0/0	100/100	100.00/100	1.4	15.3
DI	68.75	26.62	0/0	100/100	100.00/100	1.4	17.8

Table 6 also presents study findings on the variability of the subscale scores and the proportion of respondents scoring at the floor and ceiling levels. The rating scale values ranged from 0 (not at all) to 4 (extremely) or 0 to 100 (transformed scores) for all subscales with the exception of IFC and WC which were reverse scored. In the reversed scored scales the values are also reversed (0 = extremely and 4 = not at all). Six out of nine subscales had a complete range of scores. The scales without the full range (FCGT, GTP and UR) had higher mean scores and lower variability. Overall, the subscale scores evidenced fairly substantial variability.

Ceiling and floor effects demonstrated that most scales had a full range of scores. Specifically, the findings indicate that seven of the nine subscales had less than or equal to 1.5% of respondents at the floor. In general, there was a higher percent of respondents scoring at the ceiling than the floor, the exceptions again were the scales with reverse scoring (i.e., 4 = not at all; 0 = extremely). Ceiling effects were highest for the UR scale which is expected given its importance for behavioral change that is required based on one's HNPCC status (i.e., carrier versus non-carrier).

Reliability and Validity of HD-GT Scale

The reliability and validity of the subscales of the HD-GT were examined in the current study. Cronbach's alpha coefficient was used to assess internal consistency. Correlations among the subscales are useful preliminary measures of the construct validity of the entire scale. These findings are summarized in Table 7.

Table 7. HD-GT – Correlations and Internal Consistency¹ (N = 75)

Scale	IFC	FCGT	GTP	WC	SGTR	UR	TB	CGL	DI
IFC	(.89)								
FCGT	-.37 ^b	(.81)							
GTP	-.41 ^b	.55 ^a	(.91)						
WC	.45 ^a	-.49 ^a	-.45 ^a	(.86)					
SGTR	-.42 ^a	.52 ^a	.59 ^a	-.54 ^a	(.83)				
UR	-.24 ^c	.38 ^b	.58 ^a	-.31 ^b	.37 ^b	(.77)			
TB	-.57 ^a	.35 ^b	.13	-.48 ^a	.36 ^b	.13	(.70)		
CGL	-.58 ^a	.44 ^a	.33 ^b	-.39 ^b	.32 ^b	.25 ^c	.38 ^b	(.87)	
DI	-.25 ^c	.26 ^c	.33 ^b	-.55 ^a	.50 ^a	.17	.27 ^c	.11 ^c	(.78)

Note: IFC = Impact of Familial Cancer; FCGT = Family Challenges Genetic Testing; GTP = Genetic Testing Preparation; WC = Wait-Time Concerns; SGTR = Support for Genetic Testing Results; UR = Understanding Risk; TB = Transmission Beliefs; CGL = Communication around Genetic Link; DI = Disclosure Issues.

¹Cronbach's alpha coefficient is bracketed in the diagonal.

^a < .001; ^b < .01; ^c < .05

Alpha coefficients for the nine subscales were at or above recommended levels, ranging from a low of .70 for the TB subscale to a high of .91 for the GTP subscale. These findings suggest that all of the subscales have good internal consistency.

The magnitude and significance of correlations among the components of a scale are used to denote the distinctiveness of each component. At the first step, consideration is given to the pattern of correlations among the subscales. At the second step, the focus shifts to the comparability of the size of correlations among scales to each subscale's internal consistency.

The subscales of the HD-GT depict significant low to moderate, positive correlations with each other except for IFC and WC. The subscales IFC and WC negatively correlate with all other subscales as expected for reverse scored scales. That is, lower impact of familial cancer and less wait-time emotional challenges are associated with greater importance given to genetic testing and being emotionally and cognitively prepared, being supported by family members and health care providers while receiving results, understanding the positive implications of regular screening and healthy living, being aware of the profile of cancer in the family, having all at-risk family members open to and accepting of HNPCC information, and encountering few difficulties in telling others about HNPCC risks.

One final noteworthy point relates to the size or magnitude of the correlation between two subscales in comparison to the internal consistency value for each of them. As indicated by the findings in Table 7, all of the alpha coefficients are much larger than any Pearson's r value. This finding along with the significant inter-correlations noted

above support the premise that each subscale is measuring something unique and therefore makes a distinct contribution to the overall HD-GT scale.

Discussion

This pilot study was designed to collect baseline data on the HD-GT for the purpose of conducting a preliminary examination of the psychometric properties of this scale. Study findings provide strong support for the high quality of the data collected from the targeted population. That is, there was minimal missing data and an acceptable use of response choices comprising the rating scale for most items. As well, the findings provide good support for the item-level scaling assumptions. The variations noted in certain subscale means and deviations were expected for the most part and can be attributed to item content deviations designed to capture the full range of the variable being measured.

Study findings also provide strong support for item-scale assumptions. The item internal consistency was met with all items to hypothesized scale correlations $>.40$ with one exception. As well, most item-scale correlations fell within the $.40$ to $.70$ range with some notable exceptions, providing general support for the item-scale correlation assumption. Finally, the findings provide good support for the item discriminant validity assumption. That is, most items evidenced stronger correlations with their hypothesized subscales than other subscales comprising the HD-GT scale. It is anticipated that a larger sample size within the larger study will address some of the concerns around the equality of item-scale correlation and item discriminant validity assumptions.

With regard to scale properties, the distribution of subscale scores followed a logical pattern based on theoretical suppositions, evidenced good variability, and had acceptable floor and ceiling effects. As well, reliability findings provide strong support for the internal consistency of each subscale. Finally, the statistically, significant low to mid-range correlations among the subscales and their lesser value than corresponding alpha coefficients provide tentative support for the subscales distinctiveness and construct validity.

CHAPTER 5

Discussion

The current pilot study examined the psychometric properties of the newly developed HD-GT scale. This chapter begins with a discussion of the psychometric properties of the HD-GT in relation to the literature and is followed by a discussion of the content of the subscales. Where applicable, findings are compared and contrasted to the existing literature.

Psychometric Properties

The approach to scale development and testing presented in Chapter 4 was based on the work of Ware and Gandek (1998). Other researchers have used the methodological approach suggested by these authors (Radwin, Alster, & Rubin, 2003; Radwin, Wascho, Suchy, & Tyman, 2005). It appears that this is a very acceptable method for evaluating the psychometric properties of scales.

Sample and Target Population

The target population was individuals from high and intermediate risk families registered in the Provincial Medical Genetics Program of Newfoundland and Labrador who had completed predictive DNA testing for HNPCC. The personal characteristics of pilot study respondents were similar to those of an earlier quantitative study based on a

sample derived from this registry (Way et al., 2008). Both samples appear to be representative of the target population.

For both the earlier quantitative study and the current study, the majority of respondents were female, HNPCC carriers, and unaffected by cancer. Colon cancer was the dominant type for those individuals who reported having cancer. The mean ages and age range were also similar in the two studies. Survey respondents in both studies were primarily of the intron 5 splice site mutation type. The vast majority of respondents in both studies were actively involved in cancer screening.

Scale Administration

Previous experience with this population had suggested that there could be issues with a self-administration approach to data collection (Way et al., 2008). Based on this knowledge, the research team decided that the respondents should be given a choice of face-to-face completion with an interviewer, telephone administration with an interviewer, or self-administration of the scale. All three options were utilized and data completeness was similar, regardless of the presence or absence of interviewer support. It can therefore be concluded that this scale is appropriate for utilization under variant conditions (Polit & Beck, 2008; Streiner & Norman, 2003).

Subscale Structure

The subscale structures were created based on a number of methods. Initially, exploratory factor analysis was utilized but failed to produce factors that supported the constructs of the substantive theory. This led to the generation of a correlation matrix for

all of the subscale items in which each item was analyzed for the strength and significance of its correlation with all other items. This approach allows a number of assumptions to be examined concurrently such as item internal consistency, equality of item-scale correlation and item discriminant validity (Ware & Gandek, 1998). These assumptions help establish the suitability of including an item in a particular scale (Ware & Gandek). In addition, conceptual logic was utilized for inclusion of some items based on the constructs of the substantive theory. This concept is supported by several authors, especially given the relatively small sample size for the pilot (Radwin et al., 2003, 2005; Ware & Gandek). The result of these methods was that the HD-GT scale has nine subscales containing 52 items.

Data Quality and Item-Level Scaling Assumptions

Ware and Gandek (1998) summarize key steps to follow for testing data quality and item-level scaling assumptions. Data quality involves examining the extent of missing data combined with the frequency distribution or spread of scores across the response choices of a rating scale. Item-level scaling assumptions refer to the pattern observed in the spread of item means and standard deviations within each individual subscale.

In accordance with the guidelines for data quality, a large amount of missing data for specific items may be indicative of understanding and/or interpreting problems. The current study evidenced minimal missing data with most respondents having complete data for all of the HD-GT subscales. This low percentage of missing data can be attributed to a number of factors. First, many of the respondents had been living in HNPCC families for quite a while and had participated in related research. Thus,

familiarity with the terminology would have facilitated understanding and interpretation of the statements. Second, the readability of the scales was at an acceptable grade level (less than 10) for survey research (Readability Formulas, n.d.; Streiner & Norman, 2003).

Study findings also indicated that respondents had minimal issues with interpreting the response choices given that all of the steps (anchors) were used for most items.

According to Ware and Gandek (1998), this finding indicates good data quality.

With regard to item-level scaling assumptions, the proposed guidelines suggest that items means and standard deviations should be fairly comparable within a given subscale (Ware & Gandek, 1998). This assumption held for most subscales of the HD-GT. Item means and corresponding standard deviations demonstrating the greatest difference from subscale norms could be due to expected variations in individual level emotional responses to targeted content areas. Ware and Gandek note that this is an acceptable level of deviation for items dealing with emotional content and/or perceived support needs.

Scale Level Assumptions

Item internal consistency, equality of item-scale correlations and item discriminant validity were the Likert scaling assumptions used to assess the HD-GT. The findings suggest that the subscales of the HD-GT fulfill the criteria for the Likert scaling assumptions.

Item internal consistency, for the most part, fell within the proposed guidelines by Ware and Gandek (1998). Those items that fell below the selected $\geq .40$ cutoff for inclusion in a particular subscale, and/or evidenced higher correlations with related subscales, were retained because they were needed to strengthen the overall content

validity of the specified subscale. This is an acceptable practice in the early stages of scale development (Ware & Gandek).

With regard to equality of item-scale correlations, there were several exceptions to the requirement that each item contribute approximately equal amounts to the total subscale. The items in question were those addressing emotional content which may have been responsible for the observed discrepancies. For example, items 14 and 16 of the IFC subscale deal with the stress associated with family cancer patterns, suffering and early deaths. The decision was made to keep these and other items because of their important theoretical content. According to the literature, families with hereditary cancers experience conflicting emotions as they struggle with multiple losses and search for meaning and understanding of causal factors involved with the disease (Carlsson & Nilbert, 2007; Codori et al., 2001; Jarvinen et al., 2000; Lerman et al., 1996).

Finally, the item discriminant validity assumption held for most subscales of the HD-GT. On a few occasions, a particular item did evidence equal to or greater than correlations with subscales other than the one intended. However, the internal consistency values for the subscales in question were very good to excellent and the number of scale items acceptable. One possible factor responsible for the observed deviations from this assumption is the study's small sample size. As Ware and Gandek (1998) note more vigorous testing in similar and/or larger samples is needed to finalize item inclusion or exclusion. Thus, it would be premature with a pilot study to remove questionable items.

Scale Level Descriptive Statistics

Ware and Gandek (1998) recommend examining summated rating scales for equivalency of mean and standard deviation scores within comparable samples. A second important consideration for subscale scores is the observed pattern across the response choices (floor and ceiling effects).

When subscale scores were assessed for approximate equivalency by using meaningful pilot sample subdivisions (carrier status, cancer presence, gender and age), some differences were observed between carriers and non-carriers of HNPCC. It could be argued that these findings are due to the variability in the psycho-emotional challenges facing the two groups. More importantly, despite the statistical differences in certain scores, the two groups paralleled each other in terms of general understanding, attitude and feeling levels. From this perspective, a tentative conclusion from the pilot study is that the observed differences are not of a substantial nature. This finding supports previous findings by Heshka et al. (2008). These authors found in a systematic review of existing studies that there was no significant impact on psychological outcomes in either carriers or non-carriers as a result of genetic testing. This study also reported no significant differences in risk perceptions of both groups at 12 months. Meiser (2005) also reported that the studies on HNPCC groups consistently found that carriers had only temporary increases in generalized anxiety immediately following disclosure of their test results and that non-carriers have no adverse long-term effects as a result of having genetic testing.

Reliability and Validity of the HD-GT Subscales

Following construction of the subscales and examination of their adherence to item-level and scale-level assumptions, consideration was given to reliability and validity issues. Preliminary findings indicate that the subscales of the HD-GT are both valid and reliable.

Cronbach's alpha coefficient was used to assess the internal consistency of each subscale. The alpha coefficients for the nine subscales were found to be at or above recommended levels. These preliminary findings suggest that all of the subscales have good internal consistency (Polit & Beck, 2008).

Calculation of inter-correlations among the subscales is recommended as an initial step in assessing the construct validity of the entire scale. The subscales of the HD-GT have significant low to moderate, positive correlations with each other, except for those reverse scored (IFC and WC). A major premise of the model from which the HD-GT was developed is that living in families with a strong history of hereditary cancer (construct one) influences how well individuals accept the hereditary link to cancer and are motivated to become involved in genetic testing. It is argued that this context is important because it influences the individual's thought processes and impacts integration of and adjustment to the facts on emotional and behavioral levels (Way et al., 2008). Therefore, it was expected that the subscales of the HD-GT would correlate well with each other.

Interpretation of Subscale Scores

The various subscales developed and tested in this pilot study compare favorably with what is already reported in the literature and also provide new insights into the

psychosocial and behavioral impact of genetic testing on individuals and families with HNPCC. By developing the HD-GT from a qualitative data base, the content is steeped in the personal experiences of individuals from HNPCC families. Various authors argue that instrument item-content generated from qualitative data is more likely to capture the experiences of targeted groups (Coyle & Williams, 2000; McAllister, 2001). It is also argued that clinical tools developed in this manner have better content and face validity and excellent psychometric properties (Gilgun, 2004). A discussion of the HD-GT subscales in terms of what they were designed to measure and how this relates to previous research findings in this area is provided in the following paragraphs.

The HD-GT scale is designed to measure two constructs, living in families with a strong history of hereditary cancer, and becoming aware of genetic testing and living the process, of the substantive theory, *Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases*. The construct, living in families with a strong history of hereditary cancer, is primarily measured by the first subscale which assesses the impact of a family history of cancer on genetic testing decision-making (IFC).

The low scores observed with the IFC suggest that the family's history of cancer has a significant impact on genetic testing decision-making. This finding supports previous research findings on the importance of a pervasive cancer presence for motivating people to seek genetic testing (Bleiker et al., 2003; d' Agincourt-Canning, 2005; Hadley et al., 2003; McAllister, 2002, 2003; Pieterse et al., 2005). Study findings also suggest that the experience of living in these families shape individuals' understanding of personal cancer risk. Support for this finding is also found in the literature (Bleiker et al., 2003; Codori et al., 1999; d' Agincourt-Canning, 2005; Hadley

et al., 2003; McAllister, 2002, 2003; Pieterse et al., 2005). Finally, several researchers report that individuals within high-risk families commonly overestimate their risk for developing the disease (Balmana et al., 2004; Bleiker et al., 2003; Braithwaite et al., 2004; McAllister, 2002).

The second construct, becoming aware of genetic testing and living the process, is measured by eight subscales of the HD-GT. The first subscale measuring this construct (FCGT) deals with the challenges faced by family members in decision-making around genetic testing. The high scores for this subscale suggest that survey respondents placed high value on having all potentially at-risk family members participate in genetic testing but were challenged by facilitating their acceptance of it. The items focusing on the importance of genetic testing for everyone presumed to be at high risk evidenced high mean scores and low variances. This finding concurs with what has been reported in the literature on how families influence members' awareness of cancer risk and decisions about becoming involved in genetic testing (Bleiker et al., 2003; Gaff et al., 2005; Koehly et al., 2003; Peterson et al., 2003).

Two of the subscales address preparation for and involvement in the process (GTP) and wait-time concerns (WC). The moderate to high scores in the GTP subscale suggest that respondents value being adequately emotionally prepared for genetic testing and having the appropriate information. Although only a few research studies address this, several authors suggest greater research efforts are needed to inform genetic counselors about the needs and concerns of individuals and families seeking genetic testing (Bleiker et al., 2003; Pieterse et al., 2005). The other important finding of the current study is that not everyone needs support from a genetic counselor throughout the process. This finding

concur with the conclusions derived from a review of the literature by Bleiker et al. These authors emphasize the need for further research to determine the professional support needs of individuals and families.

The moderate scores in the WC subscale indicate that study respondents experienced some level of emotional difficulty while waiting for genetic testing results. This finding is comparable to Meiser's (2005) conclusions following an extensive review of relevant literature. However, as Meiser noted, the limited information on the extent of psychological distress experienced by individuals at this time is an important gap in our understanding.

Additional subscales are designed to capture the support needs of individuals during and following receipt of genetic testing results (SGTR) and their understanding of personal risks (UR). The moderate score range for the SGTR suggests that not everyone has high support needs prior to and during the receipt of results. Nevertheless, most respondents placed high value on receiving a follow-up letter detailing their genetic testing results. These findings reinforce the assertions made by other researchers that genetic counseling sessions should be individualized and consider family dynamics (Bleiker et al., 2003; Pieterse et al., 2005).

The relatively high UR subscale scores in the current study indicate that most respondents understood the importance attached to being proactive in attitude and action towards healthy living and screening behaviors. According to McAllister (2002), previous quantitative studies have failed to consistently predict outcomes from genetic testing. In fact, most of the literature has focused on the psychological impact of genetic testing, as opposed to its impact on subsequent behavior. Most importantly, it cannot be

assumed that individuals will adopt appropriate screening behaviors as they become aware of their risk status (Meiser, 2005). So while respondents may understand their risk and intend to participate in recommended screening, previous study findings suggest that screening intentions do not always translate into actual behavior (Bleiker et al., 2005). In fact, Bleiker et al. found that while noncompliance was rare, there were instances of significant delays in the screening practices.

Another subscale is designed to capture family members' awareness of and beliefs about cancer transmission (TB) within families. The moderately high score range for this subscale could be a function of actual variations in cancer patterns or awareness levels. One exception to this general statement is that most respondents perceived increases in cancer among young people. This perception is supported by actual statistics from local, national and international contexts (Lynch & de la Chapelle, 2003; Lynch et al., 2008; Merg et al., 2005; Stuckless et al., 2007).

The remaining subscales are developed to measure the importance attached to and the perceived receptivity of individuals to information about HNPCC family risk (CGL) and issues confronting them while disclosing this information to others (DI). The moderately high scores for the CGL suggest that most family members, young and old alike, wanted information about HNPCC and were perceived to understand it. While the research is fairly extensive in terms of how information is communicated to family members, there are limited research findings reported in the literature on how well this information is received and understood (Gaff et al., 2005; Peterson et al., 2003).

In contrast, the moderately high scores for the DI suggest that respondents may have encountered some difficulties in communicating this information to other family

members. This finding is comparable to what others have reported about disclosure difficulties (Carlsson & Nilbert, 2007; Espen et al., 2007). Communication issues can have significant implications for how other family members understand their risk, whether or not they seek genetic counseling and testing and if they engage in screening (Gaff et al., 2005; Koehly et al., 2003; Peterson et al., 2003). Several authors also suggest that certain individuals could benefit from ongoing contact with geneticists to receive up-to-date information about HNPCC and its management, as well as, for support in the disclosure of information to other family members (Collins et al., 2007; Gaff et al., 2005; Koehly et al., 2003; Mesters et al., 2005, Peterson et al., 2003).

Summary

Preliminary findings from this pilot study indicate that the HD-GT is a psychometrically sound scale. The use of qualitative data to construct the HD-GT has resulted in an instrument that is steeped in the experiences of individuals living in families with HNPCC. This instrument has been developed based on the substantive theory Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases and the content and results provide support for other findings reported in the literature. In conclusion, the preliminary findings also indicate that the subscales are appropriate for use under variant conditions and appear to be sensitive enough to measure the wide-range of psychosocial and behavioral implications of genetic testing.

CHAPTER 6

Limitations and Implications

This chapter presents a discussion on the limitations and implications of the pilot study findings. The first section summarizes the limitations and strengths of the study. The second section presents an overview of the implications for nursing practice, education, leadership/administration, and research.

Limitations and Strengths

The small sample size in this study limits the generalizability of the findings, and results should be interpreted with caution. Despite the good response rate (63%), the study is based on the responses of only 45 carriers and 30 non-carriers. It is hoped that the ongoing larger study with a greater sample size will validate the findings of this pilot study. In addition, there may be some response bias as the sample population has previously been involved in other studies relating to HNPCC and some respondents may have given answers that they thought were expected.

One of the strengths of this study has been the development of a monitoring scale that is easily understood and can be administered with or without support of a professional. This will allow the scale to be utilized more broadly in practice. A second strength is that it was developed from qualitative data derived from a study conducted locally and, therefore, the content would be expected to be quite relevant to the HNPCC population in this province.

Implications

Study findings have important implications for nursing practice, education, leadership/administration, and research. The following discussion presents the implications for each of these components in a separate section.

Nursing Practice

It is widely documented in the literature that the family physician and other primary care practitioners must be cognizant of the features of HNPCC, be aware of the extracolonic cancers associated with HNPCC, and be able to do an extensive family history on the patient. These clinicians play an important role in the identification of high-risk individuals and provide follow-up care after a diagnosis has been made. Health care providers (HCPs) are also an important link in encouraging individuals to engage in the recommended screening (Lindor et al., 2006; Lynch et al., 2008; Merg et al., 2005). With the current shortage of nearly all HCPs, it is extremely important that nurses and nurse practitioners (NPs), working to their full scope of practice, assume some of these roles.

Nurses and NPs are in a unique position to identify patients who would benefit from genetic counseling and/or genetic testing. This may occur in the context of other services being provided such as in cervical screening clinics and wellness clinics. Nurses and NPs may have responsibility for collecting patient and family history information. According to Kurnat-Thoma (2008), a complete and thorough family history is the best screening mechanism for identifying potentially at-risk family members. This researcher proposes that the nurse's role in taking family histories, making referrals for genetic

testing, and supporting patients while they undergo screening is crucial. Since nurses and NPs work in all settings – acute care, long term care, community, education, and in private industry, they are in the unique situation of being the one HCP who typically spends the most time with a patient. This gives them the opportunity to get to know patients better than other HCPs and gather information from and provide relevant information to patients. Nurses in all settings should assess the structure of families, as well as their communication patterns and how these factors may influence decisions about genetic testing.

The Amsterdam I, Amsterdam II and Bethesda guidelines (Appendix A) can assist nurses in assessing which patients are appropriate candidates for genetic counseling. Given the importance of clinical judgment, especially in cases where there is suspicion of HNPCC but genetic testing has been inconclusive, nurses must know the cardinal signs of HNPCC and also be able to identify potential patients based on pattern recognition and family history as recommended by Merg et al. (2005). This can be especially important in small families or in cases where there is reduced penetrance.

The screening adherence rates among these high-risk, HNPCC families tend to be below recommended levels and there are often a number of individuals at risk in the family (Lindor et al., 2006). Nurses and NPs must be made aware of appropriate and evidence-based screening guidelines and recommendations. These recommendations are vitally important for individuals who are mutation positive or for whom the genetic testing is inconclusive. Nurses and NPs can also provide assistance by facilitating communication among family members as well as educating individuals and families about screening.

In addition to educating clients and making the necessary referrals, nurses and NPs can help in assessing and meeting physical, psychological and social needs of individuals in these families. They are also equipped to be coordinators of care for the patient, who sometimes finds it difficult to navigate the health care system.

Many authors promote a multidisciplinary approach to genetic services for patients with HNPCC (Claes et al., 2005; Collins et al., 2007; Kurnat-Thoma, 2008; Meiser, 2005). Nurses have an important role to play in this essential multidisciplinary collaboration. The nurse's role can include obtaining thorough family histories, promotion of recommended screening and health maintenance practices, supporting individuals undergoing screening, as well as making referrals for genetic counseling and testing (Kurnat-Thoma).

Unfortunately to date, Canadian nursing organizations have not identified genetics to be within the scope of professional nursing practice. It appears that nurses in the United States have taken more of a leadership role in developing roles for nurses in the field of genetics; however nurses in Canada do deliver genetic services (Bottorff et al., 2005). Some of the roles currently held by Canadian nurses in this field include genetic counseling, community genetics, genetics education, clinic coordination and genetics research (Bottorff et al.). This is clearly an area where further development is needed to define the required competencies for nurses to provide this type of service.

Nursing Education

The results of this study have important implications for basic nursing education and continuing education for nurses and NPs. Nurse educators must be aware of the role

for nurses in identifying patients who could benefit from genetic counseling as well as in recommending evidence-based cancer surveillance and be prepared to teach these competencies and skills. In addition, nurse educators must teach their students the skills for assessing and helping meet patients' psychological needs, assessing family structure, as well as strategies for facilitating individual and family discussions and decision-making around genetic counseling and testing. In addition, students need to be aware that coordination of care is an integral part of the nurses' role and that it has direct implications for the well-being of the patient. Furthermore, the role of the nurse on multidisciplinary teams should be a component of basic nursing education.

Continuing education for nurses and NPs should include education on HNPCC and its cardinal features, such as incomplete penetrance, extracolonic cancers, early age of onset, etc., so that nurses can assist in identifying potential patients and families who could benefit from genetic counseling and testing. NPs, in their role as primary care practitioners, must be aware of the importance of taking an extensive and appropriate family history so that the potential presence of HNPCC can be detected. Continuing education programs should provide evidence-based recommendations for cancer surveillance as well, so that nurses and NPs can encourage and support their patients to engage in recommended screening protocols. As well, education should include information about the psychosocial and behavioral implications that arise from being part of a HNPCC family.

In addition, as noted in a previous section, once competencies have been defined for nurses in genetics, they must be taught in nursing education at both the basic and the continuing education levels. There may also be a role for graduate programs in which

there is an opportunity for nurses to have designation as a clinical nurse specialist in genetics. Finally, certification programs may need to be developed in this specialized area.

Nursing Leadership and Administration

Nursing administrators are constantly being challenged to find alternate methods of service delivery and to operate within government- allocated budgets. Having all nurses work to their full scope of practice is one method that is currently on the forefront as a strategy to help manage the shortage of all HCPs in the system. It has been identified in the preceding section that there is a role for nurses to play in caring for and/or coordinating the care of patients and families with HNPCC. Nurse administrators would be wise to pursue these models in the current health care environment.

Given the need for competencies to be developed to incorporate genetics into the scope of professional nursing practice, nursing leaders and administrators in all nursing organizations must promote this requirement and engage in appropriate policy development to ensure this option materializes. A strategic approach will be needed at all levels of nursing leadership and administration in order to bring this need into existence.

Nursing Research

This study utilized the findings from a qualitative study to design an instrument to measure the psychosocial and behavioral impact of genetic testing for HNPCC. This pilot study is part of a larger study with longitudinal components which is ongoing and is using a descriptive correlational design to develop, validate and evaluate monitoring tools. It is

anticipated that the longitudinal component of the larger study will further validate the HD-GT and allow generalizability of the preliminary findings of this pilot study.

Research is ongoing in a larger study to further test the substantive theory, *Confronting and Accepting the Challenges of Living in Families with Genetic-Linked Diseases* (Way et al., 2008). Only the first two constructs of this theory have been utilized in the development of the HD-GT scale. Another scale is also being developed and tested from the data pertaining to the third construct, struggling to adjust. This scale will measure how the individuals and families adjust to their mutation status and potential diagnosis, including adhering to recommended screening protocols. The combination of these two scales will measure the complete psychosocial and behavioral adjustment of living with HNPCC. It is hoped that the larger study will also demonstrate that these tools can be generalized for use with individuals and families with other genetic-based diseases.

While there have been significant advances in DNA technology in the past number of years, there is still a group of patients for whom this technology has not been successful. As indicated in Chapter 1 of this thesis, only approximately one half of HNPCC families will have a MMR mutation identified. It is presumed that there are alterations not yet detected or genes involved that have not yet been discovered. In addition, some individuals and families who have been presumed to be negative may have hidden alterations in known predisposition genes. This indicates a need for further research in the field of genetics. In addition, there are implications for nursing research into the psychological implications of this inconclusive finding.

Conclusion

This pilot study tested the psychometric properties of the HD-GT scale. This scale was developed from data from a previous qualitative study and is meant to measure the psychosocial, behavioral and emotional impact of genetic testing for HNPCC.

Preliminary findings from this component of the project were able to confirm that this scale was appropriate to use under variant conditions (face-to-face, telephone and self-administered). In addition, subscale and overall scale structure were validated and the scale met Likert scaling assumptions.

Although these findings cannot be generalized at present, it represents work that has not previously been done with this population and offers the opportunity for further research. The larger study that is ongoing will allow for this research and for further refinement of the scale.

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APPENDIX A**Criteria for Diagnosis of HNPCC**

Amsterdam I criteria

At least three relatives have histologically verified colorectal cancer (CRC) and all of the following criteria are met:

- One individual is a first-degree relative of the other two;
- Disease is present in at least two successive generations;
- At least one of the relatives with CRC is diagnosed at <50 years of age;
- Familial adenomatous polyposis (FAP) has been eliminated as a possibility.

Amsterdam II criteria

At least three relatives have some form of hereditary nonpolyposis colorectal cancer (HNPCC) - colorectal cancer, endometrial, stomach, ovary, ureter/renal pelvis, brain, small bowel, hepatobiliary tract, and skin (sebaceous tumors):

- One is a first-degree relative of the other two;
- At least two successive generations affected;
- At least one of the HNPCC cancers should be diagnosed at <50 years of age;
- FAP is excluded as a possibility in any CRC cases;
- Tumors are verified whenever possible.

Bethesda Guidelines for testing of colorectal tumors for microsatellite instability

- Individuals with cancer in families that meet the Amsterdam Criteria;
- Individuals with two HNPCC-related cancers, including synchronous and metachronous colorectal cancers (CRC) or associated extracolonic cancers;^a

- Individuals with CRC who have a first-degree relative with CRC and/or HNPCC-related extracolonic cancer and/or a colorectal adenoma. One of these cancers is diagnosed less than 45 years of age, and the adenoma is diagnosed less than 40 years of age;
- Individuals diagnosed with CRC or endometrial cancer less than 45 years of age;
- Individuals with right-sided CRC that evidences an undifferentiated pattern (solid/cribiform) on histopathology and are diagnosed less than 40 years of age; ^b
- Individuals with signet-ring-cell-type CRC diagnosed less than 45 years of age; ^c
- Individuals with adenomas diagnosed less than 40 years of age.

^a Endometrial, ovarian, gastric, hepatobiliary, or small-bowel cancer or transitional cell carcinoma of the renal pelvis or ureter.

^b Solid/cribiform is defined as poorly differentiated or undifferentiated carcinoma composed of irregular, solid sheets of large eosinophilic cells and containing small gland-like spaces.

^c Composed of more than 50% of signet ring cells.

Note: Derived from Merg, Lynch, Lynch and Howe (2005)

APPENDIX B**Ethical Approval**



Faculty of Medicine

Human Investigation Committee
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February 12, 2007

Reference #08.19

Dr. Christine Way
School of Nursing
Health Science Centre

Dear Dr. Way:

This will acknowledge your correspondence dated, February 11, 2008, wherein you clarify issues and provide a revised consent form, cover letter and telephone survey script for your research study entitled **"Psychometric testing of scales designed to monitor the psychosocial and behavioral impact of genetic testing for hereditary nonpolyposis colorectal cancer (HNPCC)"**.

At the meeting held on January 31, 2008, the Human Investigation Committee (HIC) agreed that the response and revised consent form could be reviewed by the Co-Chairs and, if found acceptable, full approval of the study be granted.

The Co-Chairs of the HIC reviewed your correspondence, approved the revised consent form, cover letter and telephone survey script and, under the direction of the Committee, granted **full approval** of your research study. This will be reported to the full Human Investigation Committee, for their information at the meeting scheduled for February 14, 2008.

Full approval has been granted for one year. You will be contacted to complete the annual form update approximately 8 weeks before the approval will lapse on **January 31, 2009**. It is your responsibility to ensure that the renewal form is forwarded to the HIC office not less than 30 days prior to the renewal date for review and approval to continue the study. The annual renewal form can be downloaded from the HIC website
<http://www.med.mun.ca/hic/downloads/Annual%20Update%20Form.doc>.

The Human Investigation Committee advises THAT IF YOU DO NOT return the completed annual update form prior to or on the aforementioned date of renewal;

- * Your ethics approval will lapse
- * You will be required to stop research activity
- * You will not be permitted to restart the study until you reapply for and receive approval to undertake the study again

In addition, the Human Investigation Committee will inform the appropriate authorities. To ensure proper action is taken, the appropriate officials will be notified to terminate funding.

Modifications of the protocol/consent are not permitted without prior approval from the Human Investigation Committee. Implementing changes in the protocol/consent without HIC approval may result in the approval of your research study being revoked, necessitating cessation of all related research activity. Request for modification to the protocol/consent must be outlined on an amendment form (available on the HIC website) and submitted to the HIC for review.

For a hospital-based study, it is your responsibility to seek the necessary approval from Eastern Health and/or other hospital boards as appropriate.

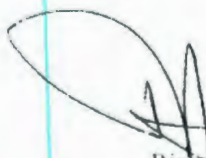
This Research Ethics Board (the HIC) has reviewed and approved the application and consent form for the study which is to be conducted by you as the qualified investigator named above at the specified study site. This approval and the views of this Research Ethics Board have been documented in writing. In addition, please be advised that the Human Investigation Committee currently operates according to the Tri-Council Policy Statement and applicable laws and regulations. The membership of this research ethics board complies with the membership requirements for research ethics boards defined in Division 5 of the Food and Drug Regulations.

Notwithstanding the approval of the HIC, the primary responsibility for the ethical conduct of the investigation remains with you.

We wish you every success with your study.

Sincerely,

John D. Harnett, MD, FRCPC
Co-Chair
Human Investigation Committee



Richard S. Neuman, PhD
Co-Chair
Human Investigation Committee

JDH:RSN/jed

C Dr. C. Loomis, Vice-President (Research), MUN
Mr. W. Miller, Senior Director, Corporate Strategy & Research, Eastern Health



Eastern Health

March 11, 2008

Dr. Christine Way
School of Nursing
MUN

Dear Dr. Way:

Your research proposal "HIC # 08.018 – *Psychometric testing of scales designed to monitor the psychosocial and behavioral impact of genetic testing for Hereditary Nonpolyposis Colorectal Cancer (HNPCC)*" was reviewed by the Research Proposals Approval Committee (RPAC) of Eastern Health at its meeting on March 7, 2008 and we are pleased to inform you that the proposal has been approved.

The approval of this project is subject to the following conditions:

- The project is conducted as outlined in the HIC approved protocol;
- Adequate funding is secured to support the project;
- In the case of Health Records, efforts will be made to accommodate requests based upon available resources. If you require access to records that cannot be accommodated, then additional fees may be levied to cover the cost;
- A progress report being provided upon request.

If you have any questions or comments, please contact Donna Bruce, Manager of the Patient Research Centre at 777-7283.

Sincerely,

Mr. Wayne Miller
Senior Director Corporate Strategy & Research
Chair, RPAC
Eastern Health

cc: Ms. Donna Bruce, Manager Patient Research Centre
Dr. Sandra LeFort, Director of MUN School of Nursing

St. Clare's Mercy Hospital
St. John's, NL
Canada A1C 5B8
T: (709) 777-5233
F: (709) 777-5272
www.easternhealth.ca

APPENDIX C**Telephone Script**

Telephone Survey Script

Study Number: _____

Location of Residence: _____

Hello. Is this 709 (____ - ____)?

If response is yes: May I speak to _____
(name)?

My name is _____ and I am calling from the Patient Research Centre in St. John's with respect to a province wide survey of HNPCC families. The Centre has initiated a number of studies with individuals who have been tested for the HNPCC gene. You may have already participated in one or more of these studies.

We are now trying to contact interested people who may be willing to help us build upon previous work. I would really appreciate some of your time this morning/afternoon/evening.

The Patient Research Centre is trying to determine individuals/families experiences with genetic testing and identify any identify any issues with screening, accessing needed resources and interacting with the health care system. Approximately 400 individuals are being contacted to answer a brief telephone survey. Your participation is very important to us. The questions will take about 45 to 60 minutes to complete. Of course, you may refuse to answer any questions. Your comments will be held in strictest confidence and no names will be used. Your participation will help us better understand the level of satisfaction with genetic testing and health care provided, as well as system changes that may be needed.

Would you mind being sent materials for you to look at more closely to help you decide about participating in the study?

Call Back Statements

If respondent is reluctant or hesitates read the following:

If you would like to speak to someone about this survey, you are welcome to call **collect** to our supervisor at 709-777-6872.

Closure

Well, that's all for now. I really appreciate your cooperation. Thank you very much for your time.

Response to "How did you get my telephone number"

If respondent is annoyed that you rang them, they may ask how you obtained their number. Respond with the following:

When you were involved in genetic testing for HNPCC, you provided the geneticist/genetic counselor with contact information (your name, telephone number and address).

If further assistance is needed

Approximately 400 individuals from different families with HNPCC will be contacted in this survey and your comments will be held in strictest confidence and no names will be used.

If necessary give them the call back statement at the top of this page.

APPENDIX D**Supporting Documents for Study Package**

1 March 2008

Dear

Thank you for agreeing to review the materials for our research study about HNPCC as discussed on the telephone. We are asking people to participate in an interview that may last about 45 to 60 minutes. A second interview might be requested in the future. These sessions will focus on your experiences with genetic testing, your reasons for choosing genetic testing and the short- and long-term impact of test results on you and your family.

Once we receive your written agreement to participate in the study, a follow-up phone call will be made by a member of our research team to schedule an acceptable time to complete the questionnaire.

Enclosed you will find a study information sheet, a copy of the survey questionnaires and two consent forms for you to review. If you require more information about the study and your participation, please contact Christine Way at 777- 6872.

We appreciate your help. We hope that the answers you provide will improve the care of individuals who have cancer in their families.

Yours sincerely,

Christine Way, PhD
Principal Investigator
NL Colorectal Cancer Study

Brief Overview of Research Study

Title: Psychometric Testing of Scales Designed to Monitor the Psychosocial and Behavioral Impact of Genetic Testing for Hereditary Nonpolyposis Colorectal Cancer (HNPCC)

Investigators: Dr. Christine Way, Dr. Mary Jane Esplen, Dr. Deborah Gregory, Dr. Patrick Parfrey, Kathy Watkins, Valerie Ludlow and Holly LeDrew

Objectives of the study:

1. To explore the usefulness of researcher-developed scales for assessing individuals experiences with genetic testing and short- and long-term adjustment in the aftermath.
2. To develop a greater understanding of the impact of genetic testing for HNPCC on carriers and non-carriers, and their families.
3. To identify relevant information that will facilitate the provision of health care services to more adequately address the needs of individuals belonging to families with documented hereditary colorectal cancer.

Rationale for the study:

Limited research has been conducted on the impact of genetic testing for individuals at risk for hereditary colorectal and related-cancers. Genetic testing provides an opportunity to help predict an individual's risk of developing these cancers. With the ability to predict or anticipate health threats, there may also be increased fear, worry or distress. Also, we know little about how such testing may influence screening and health practices. The research has tried to address these gaps by developing two scales from interview data with family members.

Brief description of the study:

The proposed study will attempt to assess individuals' experiences with genetic testing for HNPCC and adjustment to a carrier or non-carrier status. Individuals will be asked to participate in one to two interviews which are expected to last approximately 45 to 60 minutes. Interviews will focus on your experiences with genetic testing, your reasons for choosing genetic testing and the short- and long-term impact of test results on you and your family.

Procedure for obtaining consent:

A written, informed and witnessed consent will be obtained prior to the first scheduled interview.

Proposed starting date: 03/01/08

**Faculty of Medicine, Schools of Nursing and Pharmacy of Memorial
University of Newfoundland; Eastern Health; Dr. H. Bliss Murphy Cancer Centre**

Consent to Take Part in Health Research

TITLE: Psychometric Testing of Scales Designed to Monitor the Psychosocial and Behavioral Impact of Genetic Testing for Hereditary Nonpolyposis Colorectal Cancer (HNPCC)

INVESTIGATOR(S): Dr. Christine Way (709-777-6872), Dr. Mary Jane Esplen (416-340-4736), Dr. Deborah Gregory (709-729-6977), Dr. Patrick Parfrey (709-777-7261), Kathy Watkins (709-777-8142), Valerie Ludlow (709-781-0263), and Holly LeDrew (709-834-6121) (Co-investigators)

SPONSOR:

You have been invited to take part in a research study. It is up to you to decide whether to be in the study or not. Before you decide, you need to understand what the study is for, what risks you might take and what benefits you might receive. This consent form explains the study.

A member of the research team will:

- **discuss the study with you**
- **answer your questions**
- **keep confidential any information which could identify you, and**
- **be available during the study to deal with problems and answer questions**

If you decide not to take part in or to leave the study, this will not affect your normal treatment.

1. Introduction/Background:

This study is a continuation of a larger project that has been looking at the impact of hereditary cancer on people. Two of the scales that will be used were developed from information obtained from people who know their HNPCC status and are now living with this knowledge. It is hoped that the information obtained from this study will guide health care providers in giving better care to individuals and families and hopefully improve their quality of life.

2. Purpose of study:

This study is looking at the short- and long-term effects on individuals and families living with hereditary cancer, being tested for the HNPCC gene, and finding out their HNPCC status.

3. Description of the study procedures and tests:

This study will look at people's experiences with genetic testing for HNPCC and their adjustment to knowing their carrier status. You will be asked to participate in one to two interviews. The first interview might be face-to-face or by phone based upon what you would like and where you live in the province. Also, if you are willing, you may be asked to fill out the questionnaire again in the future. A copy of the questionnaires is included in your package of material.

4. Length of time:

Each phone call or face-to-face interview will take about 45 to 60 minutes.

5. Possible risks:

It is possible that during the interview you may think about difficulties to do with genetic testing and the time since then. This may cause you to have some upsetting thoughts and feelings.

You may refuse to answer any questions and end the phone call and your part in this study at any time. The interviewer may also end the phone call at any time and refer you to your genetic counsellor if he/she feels it would be helpful to you.

6. Benefits:

It is not known whether this study will benefit you.

7. Liability statement:

Signing this form gives us your consent to be in this study. It tells us that you understand the information about the research study. When you sign this form, you do not give up your legal rights. Researchers or agencies involved in this research study still have their legal and professional responsibilities.

8. Confidentiality:

Any information that you provide will be kept strictly confidential, safe in a locked file, and available only to the research team. Also, your name will not appear in any report or article as a result of this study.

9. Questions:

If you have any questions about taking part in this study, you can meet with the investigator who is in charge of the study at this institution. That person is:

Dr. Christine Way (709-777-6872)

Or you can talk to someone who is not involved with the study at all, but can advise you on your rights as a participant in a research study. This person can be reached through:

Office of the Human Investigation Committee (HIC) at 709-777-6974

Email: hic@mun.ca

Signature Page

Study title: Psychometric Testing of Scales Designed to Monitor the Psychosocial and Behavioral Impact of Genetic Testing for Hereditary Nonpolyposis Colorectal Cancer (HNPCC)

Name of principal investigator: Dr. Christine Way

To be filled out and signed by the participant:

Please check as appropriate:

I have read the consent and information sheet.	Yes { }	No { }
I have had the opportunity to ask questions/to discuss this study.	Yes { }	No { }
I have received satisfactory answers to all of my questions.	Yes { }	No { }

I have received enough information about the study.	Yes { }	No { }
---	---------	--------

I understand that I am free to withdraw from the study	Yes { }	No { }
• at any time		
• without having to give a reason		
• without affecting my future care		

I understand that it is my choice to be in the study		
and that I may not benefit.	Yes { }	No { }
I agree to take part in this study.	Yes { }	No { }

Signature of participant

Date

Signature of witness
Important for Files!

Date

To be signed by the investigator:

I have explained this study to the best of my ability. I invited questions and gave answers. I believe that the participant fully understands what is involved in being in the study, any potential risks of the study and that he or she has freely chosen to be in the study.

Signature of investigator

Date

Telephone number: _____

APPENDIX E

Survey Instruments

Demographic Profile

Study Number: _____

Today's Date: _____

We are interested in some information about your past medical history. Please use an X or write your answer in the space provided.

Gender: ___ Male ___ Female

Current Age in Years: _____

Have you ever been **screened for cancer**? ___ Yes ___ No

If **Yes**, please indicate what type of screening and how often:
(e.g., colonoscopy, every 2 years)

Type of screening	How often

Have you ever been **told you have cancer**? ___ Yes ___ No

If **Yes**, please indicate what type:

___ Bowel Cancer

___ Reproductive in women:

___ Endometrial

___ Ovarian

___ Stomach

___ Pancreas

___ Kidney

___ Other: _____

___ Other: _____

Have there been any changes in your health since the last questionnaire?

Have there been any changes in the health of close family members since the last questionnaire?

Thank you for filling out this questionnaire.

© Way, Watkins & Ludlow

HNPCC Survey Questionnaire A
(Familial Cancer and Genetic Testing Scale)

This survey questionnaire has 10 questions.

Each question has several statements that we would like you to rate from **0 (Not at all)** to **4 (Extremely)**.

Please circle the best answer for each.

Thank you.

A1. We want to know the degree to which a family history of cancer influences a person's decision to have genetic testing for HNPCC.

Using the scale given, you are asked to rate how important each statement was in helping you to decide to have genetic testing done.

0 - Not at all

1 - A little bit

2 - Moderately

3 - Quite a bit

- | | | | | | |
|---|---|---|---|---|---|
| 1. It seemed like a lot of family members were getting cancer more often and at a younger age than in past generations. | 0 | 1 | 2 | 3 | 4 |
| 2. I have many memories of close family members suffering from cancer illness and treatment effects. | 0 | 1 | 2 | 3 | 4 |
| 3. The presence of so much cancer in the family was hard to accept, and I wanted to know why. | 0 | 1 | 2 | 3 | 4 |
| 4. It was scary to see the same pattern of cancer showing up in every generation of my family. | 0 | 1 | 2 | 3 | 4 |
| 5. It was so draining to lose close relatives to cancer that it got to a point when the phone rang I wondered who was sick this time. | 0 | 1 | 2 | 3 | 4 |
| 6. With so much suffering and early deaths from cancer, I was worried about my own health and death. | 0 | 1 | 2 | 3 | 4 |
| 7. What worried me was that even when a family member seemed to beat the odds with one form of cancer, another one showed up. | 0 | 1 | 2 | 3 | 4 |
| 8. The stress of so much cancer in the family, more so in younger members, pulled some of us closer together but pushed others apart. | 0 | 1 | 2 | 3 | 4 |
| 9. I grew tired of how certain family members tried to hide the cancer from the children. | 0 | 1 | 2 | 3 | 4 |

A2. We want to know to what extent you were thinking about a genetic link to cancer in your family prior to having contact with a geneticist/genetic counsellor, and how you reacted to being told that you were potentially at risk.

Using the scale given, you are asked to rate how important each statement was in helping you to prepare for genetic testing.

0 - Not at all

1 - A little bit

2 - Moderately

3 - Quite a bit

4 - Extremely

- | | | | | | |
|--|---|---|---|---|---|
| 1. Over the years concerns were expressed by some family members that there was a cancer gene in our family. | 0 | 1 | 2 | 3 | 4 |
| 2. It was only after the geneticist made contact with family members that I really began to think that the cancer had to be a family thing and I could also be at risk. | 0 | 1 | 2 | 3 | 4 |
| 3. When I knew that there was a study looking to see if my family had the cancer gene, I was relieved. | 0 | 1 | 2 | 3 | 4 |
| 4. When I was told that there was a test that could find the cancer gene in the family, it was not a matter of "would I go for genetic testing", but "when I could have it". | 0 | 1 | 2 | 3 | 4 |
| 5. I really questioned whether knowing if I am a carrier for HNPCC would do me more harm than good (i.e., restricted insurance coverage and job prospects). | 0 | 1 | 2 | 3 | 4 |
| 6. Doing the screening for cancer every couple of years became a constant reminder of just how at risk I was by being a part of this family. | 0 | 1 | 2 | 3 | 4 |

7. Taking the prep for the colonoscopy test was such a pain that I did not want to be doing this unless it was needed.
8. With so much cancer in the family I worried that something would show up on my next screening test.

0 1 2 3 4

0 1 2 3 4

A3. Going through genetic testing may be different even for people within the same family. We want to know how informed you were and how emotionally prepared you were.

Using the scale given, you are asked to rate these statements in terms of their importance in helping you and others decide to take part in genetic testing.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

It is important to:

- | | | | | | |
|---|---|---|---|---|---|
| 1. Get enough information in a timely manner from geneticists/genetic counsellors about HNPCC. | 0 | 1 | 2 | 3 | 4 |
| 2. Receive enough information from geneticists/genetic counsellors about the genetic testing process. | 0 | 1 | 2 | 3 | 4 |
| 3. Feel no pressure to have genetic testing done. | 0 | 1 | 2 | 3 | 4 |
| 4. Know ones HNPCC status. | 0 | 1 | 2 | 3 | 4 |
| 5. Understand ones risk for HNPCC and accept the need for genetic testing. | 0 | 1 | 2 | 3 | 4 |
| 6. Know ones HNPCC status to help children/grandchildren. | 0 | 1 | 2 | 3 | 4 |
| 7. Have geneticists/genetic counsellors to support you during the genetic testing process. | 0 | 1 | 2 | 3 | 4 |
| 8. Feel support and encouragement from family and/or friends. | 0 | 1 | 2 | 3 | 4 |

A4. Certain people within families refuse to participate in genetic testing for one reason or another. Concerns were expressed about the future well-being for them and their families.

Using the scale given, you are asked to rate how well these statements describe your feelings about family members who refuse to have genetic testing.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

- | | | | | | |
|---|---|---|---|---|---|
| 1. It is important for all family members at risk for HNPCC to take part in genetic testing. | 0 | 1 | 2 | 3 | 4 |
| 2. I am really concerned about family members who refuse to go for genetic testing. | 0 | 1 | 2 | 3 | 4 |
| 3. I feel that family members who refuse to go for genetic testing do not understand their risks. | 0 | 1 | 2 | 3 | 4 |
| 4. I believe that family members who refuse genetic testing are fearful of knowing their results. | 0 | 1 | 2 | 3 | 4 |

A5. Different thoughts and emotions were experienced by individuals within and across the study families as they waited for their genetic testing results.

Using the scale given, please rate how well these statements describe your feelings about the wait time between the giving of blood for genetic testing for HNPCC and the actual receipt of your results.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

- | | | | | | |
|---|---|---|---|---|---|
| 1. It was a very trying time thinking about if I could be a carrier/non-carrier for HNPCC. | 0 | 1 | 2 | 3 | 4 |
| 2. I was not really prepared for such a long time between having the test done and getting the results. | 0 | 1 | 2 | 3 | 4 |
| 3. I was unsure about how I would receive my results (e. g., phone, letter, in person). | 0 | 1 | 2 | 3 | 4 |
| 4. I spent a lot of time thinking about how I would react to finding out my genetic testing results. | 0 | 1 | 2 | 3 | 4 |
| 5. I wondered if I would understand what my genetic testing results really meant for me. | 0 | 1 | 2 | 3 | 4 |

A6. People often report different experiences about how they receive their results for HNPCC.

Using the scale given, you are asked to rate the following statements in terms of their importance for you.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

It is important to:

- | | | | | | |
|--|---|---|---|---|---|
| 1. Have a family member and/or friend present. | 0 | 1 | 2 | 3 | 4 |
| 2. Get a phone call from geneticist/genetic counsellor prior to receiving your results. | 0 | 1 | 2 | 3 | 4 |
| 3. Have face to face contact with a geneticist/genetic counsellor when receiving your results. | 0 | 1 | 2 | 3 | 4 |
| 4. Receive a letter explaining the meaning of being a HNPCC carrier/non-carrier for yourself and others. | 0 | 1 | 2 | 3 | 4 |

A7. People react differently to receiving their genetic testing results no matter how prepared they are.

Using the scale given, you are asked to rate these statements in terms of how well they reflect your situation after getting your results.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

- | | | | | | |
|--|---|---|---|---|---|
| 1. When you know that your cancer is hereditary, you wonder and worry about when it will show up in the future. | 0 | 1 | 2 | 3 | 4 |
| 2. Despite feeling fully prepared when I went to find out my HNPCC status, I was surprised by the results. | 0 | 1 | 2 | 3 | 4 |
| 3. Knowing my HNPCC status brought a sense of closure to everything. | 0 | 1 | 2 | 3 | 4 |
| 4. The information received from the geneticist/genetic counsellor about cancer risk was very clear and useful. | 0 | 1 | 2 | 3 | 4 |
| 5. Follow-up contact with the geneticist/genetic counsellor to discuss healthy lifestyles and screening schedules would have been helpful. | 0 | 1 | 2 | 3 | 4 |
| 6. Overall, it was better to know if I was a carrier/non-carrier than not to know. | 0 | 1 | 2 | 3 | 4 |

A8. We want to know if being a carrier/non-carrier changed how you looked at healthy living and screening behaviors. What is important is how understanding of ones risk impacts choices about healthy living and screening behaviors.

Using the scale given, you are asked to rate how well these statements apply to your situation.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

- | | | | | | |
|---|---|---|---|---|---|
| 1. Encouragement and support from family and friends helps one accept the need for healthy living and cancer screening. | 0 | 1 | 2 | 3 | 4 |
| 2. Regular screening will help detect cancer at an early stage. | 0 | 1 | 2 | 3 | 4 |
| 3. Appropriate screening for HNPCC is important for timely detection of colon and related cancers. | 0 | 1 | 2 | 3 | 4 |
| 4. Early detection of cancer will help improve treatment and disease management. | 0 | 1 | 2 | 3 | 4 |
| 5. Healthy living (exercise, diet) and a positive attitude will help increase well-being and decrease stress. | 0 | 1 | 2 | 3 | 4 |
| 6. Taking responsibility for healthy living and regular screening is important. | 0 | 1 | 2 | 3 | 4 |

A9. The presence of cancer in the family is often important in helping understand ones personal risk for HNPCC.

Using the scale given, you are asked to show how well these statements reflect your family's situation.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

- | | | | | | |
|---|---|---|---|---|---|
| 1. Family members are being diagnosed with HNPCC cancers at a younger age. | 0 | 1 | 2 | 3 | 4 |
| 2. Men and women seem to have different types of first cancers. | 0 | 1 | 2 | 3 | 4 |
| 3. Different types of cancer seem to be showing up today more than in past generations. | 0 | 1 | 2 | 3 | 4 |
| 4. The number of family members with cancer seems to be greater with each generation. | 0 | 1 | 2 | 3 | 4 |
| 5. Physical appearance and personality are good signs of who will inherit the HNPCC gene. | 0 | 1 | 2 | 3 | 4 |

A10. The question 'who to tell and who not to tell about HNPCC in the family' is a concern for many people. The person who assumes the 'messenger role' may face difficulties in telling others about HNPCC in the family.

Using the scale given, you are asked to rate how well these statements apply to your situation.

- 0 - Not at all
- 1 - A little bit
- 2 - Moderately
- 3 - Quite a bit
- 4 - Extremely

- | | | | | | |
|--|---|---|---|---|---|
| 1. It is important for young family members to be told about the presence of HNPCC in the family. | 0 | 1 | 2 | 3 | 4 |
| 2. It is difficult having to tell younger family members about their possible risk for HNPCC. | 0 | 1 | 2 | 3 | 4 |
| 3. Younger family members seem to be open to information about HNPCC in the family. | 0 | 1 | 2 | 3 | 4 |
| 4. Younger family members seem to understand what HNPCC in the family could mean for them. | 0 | 1 | 2 | 3 | 4 |
| 5. It is important for all family members to be told about HNPCC. | 0 | 1 | 2 | 3 | 4 |
| 6. It is hard telling other family members about their possible risk for HNPCC. | 0 | 1 | 2 | 3 | 4 |
| 7. Family members seem to be open to information about HNPCC. | 0 | 1 | 2 | 3 | 4 |
| 8. Family members seem to understand what HNPCC could mean for them. | 0 | 1 | 2 | 3 | 4 |
| 9. It would be helpful if the messenger had guidance and support from geneticists/genetic counsellors on how to tell others about HNPCC in the family. | 0 | 1 | 2 | 3 | 4 |

10. It is important to protect the rights of others
when talking about who could be at risk for
HNPCC in the family.

0 1 2 3 4

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